chapter

19 Beyond Mendel

In this chapter

Early scientists believed that hereditary traits were located in the blood. The term "pure bloodline," which is still used today by animal breeders (**Figure 1**), is a reminder of this misconception, as is the French term Métis conferred by European fur traders on peoples of mixed Aboriginal and European "blood." Today we know that inherited traits are determined by genes, which are located along the thread-like chromosomes found in the nucleus of each cell.

The field of genetics changed quickly once scientists began to describe the location and the chemical makeup of chromosomes. Genes can now be identified and selected, and sometimes even altered. One of the most dramatic examples of changing inherited traits is the production of mice that are smarter than mice are naturally. This genetically modified strain was dubbed Doogie, after a television character who was a teenage genius.

The modification and insertion of a single gene, *NR2B*, into a chromosome of the mice improves the functioning of nerve receptors that play a key role in memory and learning. The laboratory-bred Doogie mice learn faster and remember more than normal mice. For example, scientists found that when a new and an old object were introduced into the cage with the Doogie mice, they spent most of their time exploring the new object. This indicated that they recognized and remembered the old object. Normal mice spent equal time with the new and old objects. The Doogie mice generated great excitement, because humans possess a corresponding gene.

STARTING Points

Answer these questions as best you can with your current knowledge. Then, using the concepts and skills you have learned, you will revise your answers at the end of the chapter.

1. In what part of the cell would you find genes?
2. Can you distinguish males from females by looking at their genetic material?
3. Explain how a better understanding of chromosome structure could lead to a more complete understanding of gene function.
4. Why might some people be opposed to making mice smarter?
5. Why might the research with mice prove important for people with Alzheimer's disease?

Figure 1
Animal breeders produce varieties of a species with a specific set of traits, such as these Appaloosa horses. The value of an individual animal is often determined by its bloodline, a term that dates back to early misconceptions about heredity.

Exploration — Inherited Traits

Some physical characteristics are controlled by a single gene that can be expressed in one of two ways. Try the tests below to see what phenotype you express.

- Fold your arms in front of your body.
  (a) Which arm is on top?
- Change arm position so that the other arm is on top.
  (b) Describe how it feels.
- Interlock your fingers.
  (c) Are the fingers from your left hand or your right hand on top?
- Place a strip of PTC paper on your tongue.
  (d) Could you taste the paper?
- Gather and compile the class data for all three tests.
  (e) For each test, which trait occurred most frequently in your class?
  (f) Do traits determined by dominant genes always occur with the highest frequency? Explain your answer.

19.1 Chromosomes and Genetics

Figure 1
The artist Leonardo da Vinci became interested in anatomy and dissection because of his desire to paint the human form better.

During the Middle Ages (500–1300 CE), curious individuals would sneak into caves to dissect corpses. Despite strict laws prohibiting such behaviour, the inquiring minds of early physicians and scientists compelled them to conduct their investigations. Generations of artists sketched different parts of the body (**Figure 1**), creating a guide to anatomy in the process. As a composite structure of organs began to appear, theories about function arose. The principle that structure gives clues about function also applies to genetics. However, the early geneticists had to wait for the emergence of the light microscope before investigations into genetic structure could seriously progress. The study of genes is closely connected with technology. The light microscope, the electron microscope, X-ray diffraction, and gel electrophoresis have provided a more complete picture of the mechanisms of gene action.

The discovery of the nucleus in 1831 was an important step toward understanding the structure and function of cells and the genes they contain. By 1865, the year in which Mendel published his papers, biologists knew that the egg and sperm unite to form a zygote, and it was generally accepted that factors from the egg and sperm were blended in developing the characteristics of the offspring. Even though Mendel knew nothing about meiosis or the structure or location of the hereditary material, he was able to develop theories about inheritance that adequately explain how traits are passed on from generation to generation.

At about the same time that Mendel was conducting his experiments with garden peas, new techniques in lens grinding were providing better microscopes. The improved technology helped a new branch of biology, cytology, to flourish. Cytology is the study of cell formation, structure, and function. Aided by these technological innovations, in 1882, Walter Fleming described the separation of threads within the nucleus during cell division. He called the process mitosis. In the same year, Edouard van Benden noticed that the sperm and egg cells of roundworms had two chromosomes, but the fertilized eggs had four chromosomes. By 1887, August Weisman offered the theory that a special division took place in sex cells. By explaining the reduction division now known as meiosis, Weisman added an important piece to the puzzle of heredity and provided a framework in which Mendel's work could be understood. When scientists rediscovered Mendel's experiments in 1900, the true significance of his work became apparent.

Chromosomal Theory

Learning Tip

Recall that homologous chromosomes occur in pairs and are similar in size, shape, and gene information and arrangement.

In 1902, American biologist Walter S. Sutton and German biologist Theodor Boveri independently observed that chromosomes came in pairs that segregated during meiosis. The chromosomes then formed new pairs when the egg and sperm united. The concept of paired, or homologous, chromosomes supported Mendel's explanation of inheritance based on paired factors. Today, these factors are referred to as the alleles of a gene. One factor, or allele, for each gene comes from each sex cell.

The union of two different alleles in offspring and the formation of new combinations of alleles in succeeding generations could be explained and supported by cellular evidence. The behaviour of chromosomes during gamete formation could help explain Mendel's law of segregation and law of independent assortment.

Sutton and Boveri knew that the expression of a trait, such as eye colour, was not tied to only the male or only the female sex cell. Some structures in both the sperm cell and

the egg cell must determine heredity. Sutton and Boveri deduced that Mendel's factors (alleles) must be located on the chromosomes. The fact that humans have 46 chromosomes (44 **autosomes** and 2 sex chromosomes), but thousands of different traits, led Sutton to hypothesize that each chromosome carries genes. Genes that are on the same chromosome are said to be **linked genes**.

autosome a chromosome not involved in sex determination

linked genes genes that are located on the same chromosome

The chromosomal theory of inheritance can be summarized as follows:

- Chromosomes carry genes, the units of heredity.
- Paired chromosomes segregate during meiosis. Each sex cell or gamete has half the number of chromosomes found in the somatic cells. This explains why each gamete has only one of each of the paired alleles.

As you saw in the previous chapter, chromosomes assort independently during meiosis. Each gamete receives one member from each pair of chromosomes, and each chromosome pair has no influence on the movement of any other chromosome pair. This explains why in a dihybrid cross an F_1 parent, *AaBb*, produces four types of gametes: *AB*, *aB*, *Ab*, *ab*. Each gamete appears with equal frequency due to segregation and independent assortment. Each chromosome contains many different alleles and each gene occupies a specific locus or position on a particular chromosome.

Morgan's Experiments and Sex-Linked Traits

The American Thomas Hunt Morgan was among the first of many geneticists who used the tiny fruit fly, *Drosophila melanogaster*, to study the principles of inheritance. There are several reasons why the fruit fly is an ideal subject for study. First, the fruit fly reproduces rapidly. Offspring are capable of mating shortly after leaving the egg, and females produce over 100 eggs after each mating. Female *Drosophila* can reproduce for the first time when they are only 10 to 15 days old, so it is possible to study many generations in a short period of time. Since genetics is based on probability, the large number of offspring is ideal. A second benefit arises from *Drosophila*'s small size. Many individuals can be housed in a single culture tube. A small, solid nutrient at the bottom of the test tube can maintain an entire community. The third and most important quality of *Drosophila* is that males can easily be distinguished from females. Males are smaller and have a rounded abdomen with a dark-coloured posterior segment while the larger females have a pointed abdomen with a pattern of dark bands.

Figure 2
In *Drosophila*, the allele that codes for white eyes (male fly, top photo) is recessive to the allele that codes for red eyes (female fly, bottom photo).

While examining the eye colour of a large number of *Drosophila*, Morgan noted the appearance of a white-eyed male among many red-eyed offspring (**Figure 2**). He concluded that the white-eyed trait must be a mutation. Morgan was interested in tracing the inheritance of the allele coding for white eyes, so he mated the white-eyed male with a red-eyed female. All members of the F_1 generation had red eyes. Normal Mendelian genetics indicated that the allele for red eyes was dominant. Most researchers might have stopped at that point, but Morgan did not. Pursuing further crosses and possibilities, he decided to mate two hybrids from the F_1 generation. An F_2 generation produced $\frac{3}{4}$ red eyes and $\frac{1}{4}$ white eyes, a ratio that could again be explained by Mendelian genetics. But further examination revealed that all the females had red eyes. Only the males had white eyes. Half of the males had red eyes and half had white eyes. Did this mean that the white-eyed phenotype only appears in males? Why could males express the white-eyed trait but not females? How did the pattern of inheritance differ between males and females? To find an answer, Morgan turned to cytology.

Previous researchers had stained and microscopically examined the eight chromosomes from the cells of the salivary glands of *Drosophila*. They found that females have four homologous pairs and males have only three homologous pairs. The fourth pair, which determines sex, is only partially homologous. Males were found to have one

CAREER CONNECTION

Entomologist

Entomologists study the life cycle of insects and conduct research into evolution and biodiversity. The science of entomology has made a significant contribution to understanding genetics and gene mapping. Would you like to work with fruit flies or arthropods, such as spiders and mites? Explore this field of study.

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X chromosome paired with a small, hook-shaped Y chromosome. Females have two paired X chromosomes (**Figure 3**). Since the X and Y chromosomes are not completely homologous (although they act as homologous pairs during meiosis), it was concluded that they contain different genes.

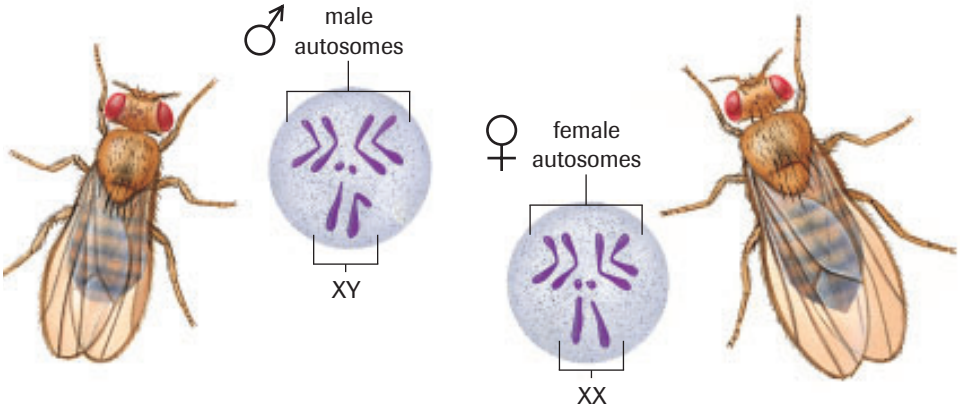

Figure 3
Drosophila contain three pairs of autosomes and a single pair of sex chromosomes.

Morgan explained the results of his experiments by concluding that the Y chromosome does not carry the gene to determine eye colour. We now know that the gene for eye colour in *Drosophila* is located on the part of the X chromosome that does not match the Y chromosome. Therefore, Morgan's conclusion was correct. The Y chromosome does not carry an allele for the eye-colour gene. Traits determined by genes located on sex chromosomes are called **sex-linked traits**.

sex-linked trait trait that is determined by genes located on the sex chromosomes

The initial problem can now be re-examined. The pure-breeding, red-eyed female can be indicated by the genotype X^RX^R and the white-eyed male by the genotype X^rY. The symbol X^R indicates that the allele for red eye is dominant and is located on the X chromosome. There is no symbol for eye colour on the Y chromosome because it does not contain an allele for the trait. A Punnett square, as shown in **Figure 4**, can be used to

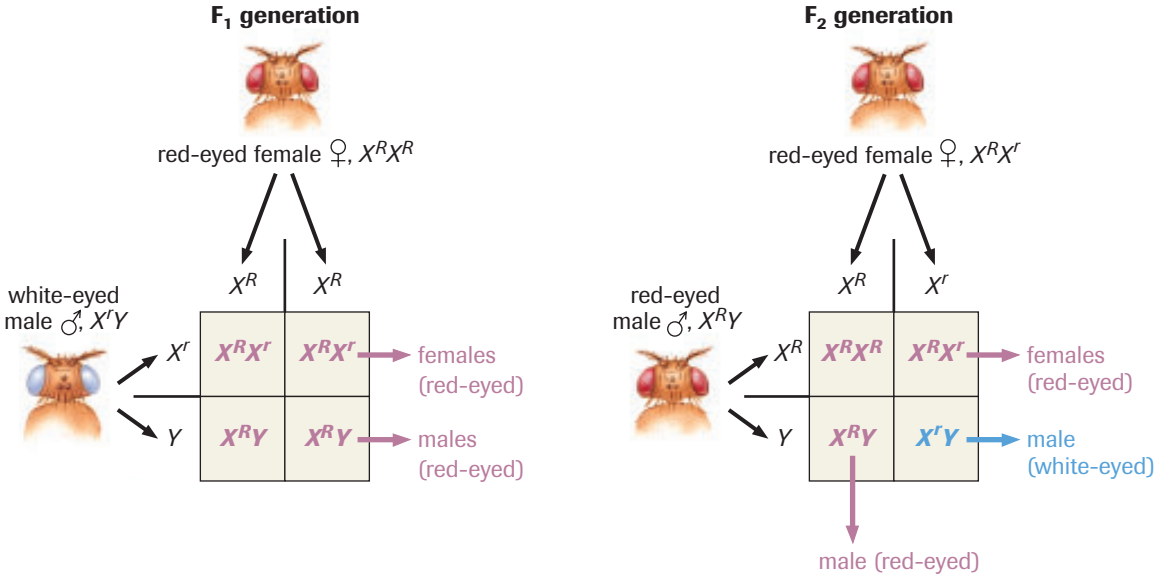

Figure 4
Punnett squares showing F_1 and F_2 generations for a cross between a homozygous red-eyed female and a white-eyed male.

determine the genotypes and the phenotypes of the offspring. All members of the F_1 generation have red eyes. The females have the genotype X^RX^r, and the males have the genotype X^RY.

The F_2 generation is determined by a cross between a male and female from the F_1 generation. Upon examination of the F_1 and F_2 generations, the question arises whether the males inherit the trait for eye colour from the mother or father. The male offspring always inherit a sex-linked trait from the mother. The father supplies the Y chromosome, which makes the offspring male.

The F_2 male *Drosophila* are X^RY and X^rY. The females are either homozygous red for eye colour, X^RX^R, or heterozygous red for eye colour, X^RX^r (**Table 1**). Although Morgan did not find any white-eyed females from his initial cross, some white-eyed females do occur in nature. For this to happen, a female with at least one allele for white eyes must be crossed with a white-eyed male. Notice that females have three possible genotypes, but males have only two. Males cannot be homozygous for an X-linked gene because they have only one X chromosome. The Y chromosome has less than 100 genes.

Recall that humans have 46 chromosomes. Females have 23 pairs of homologous chromosomes: 22 autosomes, and two X sex chromosomes. Males have 22 pairs of homologous chromosomes, and one X sex chromosome and one Y sex chromosome (**Figure 5**). It has been estimated that the human X chromosome carries between 100 and 200 different genes. The Y chromosome has less than 100 genes.

Sex-linked genes are also found in humans. For example, a recessive allele located on the X chromosome determines red–green colour-blindness. More males are colour-blind than females because females require two recessive alleles to exhibit colour-blindness. Since males have only one X chromosome, they require only one recessive allele to be colour-blind. Other sex-linked traits that affect males primarily include hemophilia, hereditary near-sightedness (myopia), and night-blindness.

This explains why **recessive lethal** X-linked disorders in humans, such as infantile spinal muscular atrophy, occur more frequently in males. This could also explain why the number of females reaching the age of 10 and beyond is greater than the number of males. Males die at birth or before the age of 10 from recessive lethal X-linked disorders.

Table 1 Possible Genotypes for *Drosophila*

Females	Males
X^RX^R	X^rY
X^RX^r	X^RY
X^rX^r	

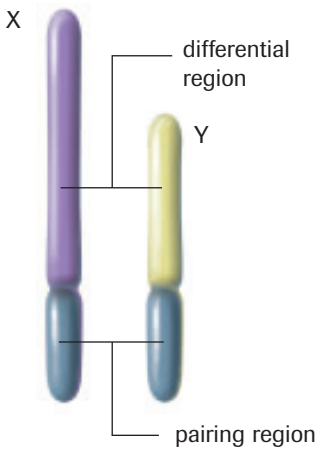

Figure 5
Sex chromosomes. Sections of the X and Y chromosomes are homologous; however, few genes are common to both chromosomes.

Barr Bodies

The difference between male and female autosomal (non-sex) cells lies within the X and Y chromosomes. Dr. Murray Barr, working at the University of Western Ontario in London, recognized a dark spot in some of the somatic cells of female mammals during the interphase of meiosis. This spot proved to be the sex chromatin, which results when one of the X chromosomes in females randomly becomes inactive in each cell. This dark spot is now called a **Barr body** in honour of its discoverer. This discovery revealed that not all female cells are identical; some cells have one X chromosome inactive, while some have the other. This means that some cells may express a certain trait while others express its alternate form, even though all cells are genetically identical. For example, if a human female is heterozygous for the skin disorder *anhidrotic ectodermal dysplasia*, she will have patches of skin that contain sweat glands and patches that do not. This mosaic of expression is typical of X chromosome activation and inactivation. In normal skin, the X chromosome with the recessive allele is inactivated and sweat glands are produced. In the afflicted skin patches, the X chromosome with the recessive allele is activated and no sweat glands are produced.

recessive lethal a trait that, when both recessive alleles are present, results in death or severe malformation of the offspring. Usually, recessive traits occur more frequently in males.

Barr body a small, dark spot of chromatin located in the nucleus of a female mammalian cell

EXTENSION

Barr Body Formation
Listen to a discussion of the formation of Barr bodies and mosaic phenotypes in females.

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LAB EXERCISE 19.A

Tracing the Hemophilia Gene

A pedigree chart provides a means of tracing the inheritance of a particular trait from parents through successive generations of offspring. Hemophilia A is a blood-clotting disorder that occurs in about one in 7000 males. The disorder is associated with a recessive gene located on the X chromosome, normally represented as X^h. Normal blood clotting is controlled by a dominant gene, X^H. The fact that a female must inherit one of the mutated alleles from her mother and another of the mutated alleles from her father helps explain why this disorder is very rare in females. Males, on the other hand, only need to inherit one recessive allele to express the disorder.

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	○ Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	○ Evidence	

Purpose

To use pedigree charts to trace the hemophilia gene from Queen Victoria

Evidence

See **Figure 6**.

Analysis

1. Study the pedigree chart of Queen Victoria and Prince Albert (**Figure 6**). Note the legend at top right.

(a) Who was Queen Victoria's father?

(b) How many children did Queen Victoria and Prince Albert have?

2. Locate Alice of Hesse and Leopold, Duke of Albany, on the pedigree chart.

(c) Using the legend, provide the genotypes of both Alice of Hesse and Leopold.

3. Locate the royal family of Russia on the pedigree chart by finding Alexandra. Alexandra, a descendant of Queen Victoria, married Nikolas II, Czar of Russia. Nikolas and Alexandra had four girls (only Anastasia is labelled), and one son, Alexis.

(d) Explain why Alexis was the only child with hemophilia.

(e) Is it possible for a female to be hemophilic? If not, explain why not. If so, identify a male and female from the pedigree chart who would be capable of producing a hemophilic, female offspring.

(f) On the basis of probability, calculate the number of Victoria's and Albert's children who would be carriers of the hemophilic trait.

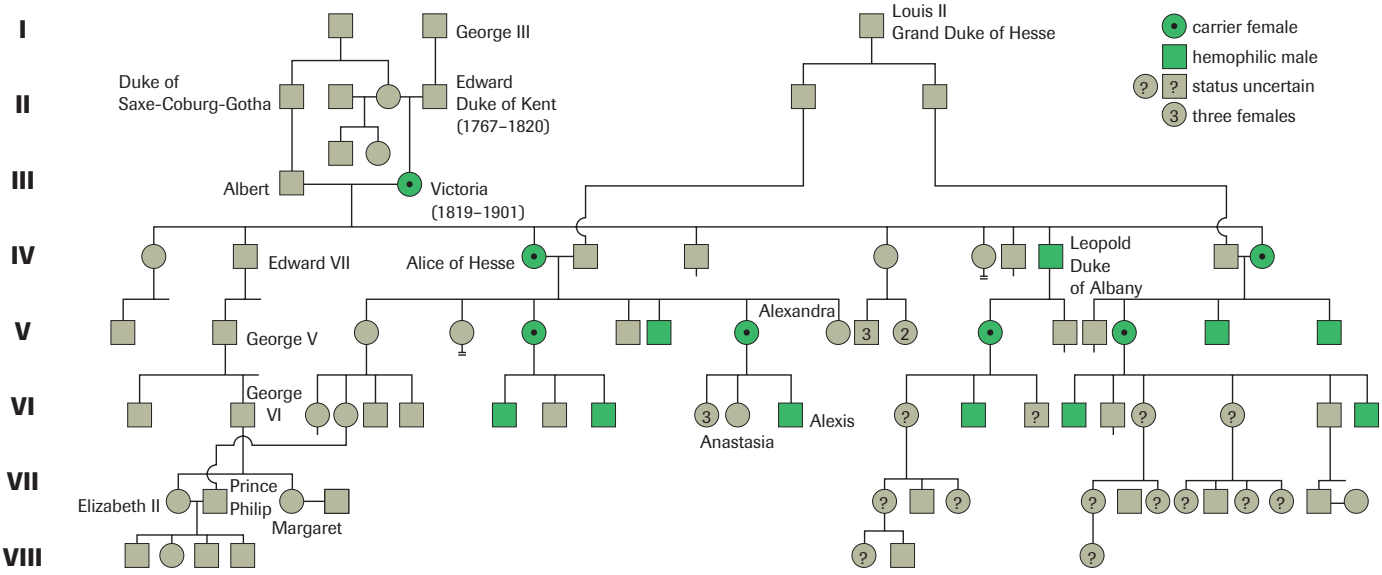

Figure 6

EXPLORE an issue

Report Checklist

- Issue
- Design
- Analysis
- Resolution
- Evidence
- Evaluation

Screening for Genetic Disorders

Screening for inherited diseases can be carried out by various methods, including detailed pedigrees and biochemical testing for known disorders. Prenatal ("before birth") diagnosis can determine the presence of many genetic conditions in the unborn fetus. Amniocentesis involves the extraction of a small sample of fluid from the amnion, the membranous sac around the fetus. Chorionic villi sampling (CVS) involves withdrawing cells from the chorion, a fluid-filled membranous sac that surrounds the amnion. CVS can yield results earlier than amniocentesis, as early as in the ninth week of pregnancy.

Before the development of a process that permitted the extraction of insulin from animals, the children of parents who passed on two copies of the recessive allele for diabetes died at a young age. Today, genetic screening can tell potential parents if they carry this allele (**Figure 7**). Huntington disease is a neurological disorder caused by a dominant allele that only begins to express itself later in life. The disease is characterized by the rapid deterioration of nerve control, eventually leading to death. Early detection of this disease by genetic screening is possible.

By having knowledge of a genetic disorder prior to birth, parents will have the opportunity to be better prepared to cope with any additional challenges the disorder may bring. Some parents may choose to terminate a pregnancy based on the results of genetic screening. This use of genetic screening is controversial.

- In small groups, research the issue of using genetic screening to detect inherited conditions. Find other ways of dealing with genetic disorders instead of genetic screening. You may wish to focus your research on one of the conditions described above.
- List the points and counterpoints against genetic screening uncovered by your group. After considering each of these, and any alternative means of dealing with genetic disorders that you found, write a statement that outlines your group's position on this issue.
- Prepare to defend your group's position in a class discussion.

Figure 7
A genetic counsellor helps a couple to assess their risks of having children with inherited diseases.

EXTENSION

The Pros and Cons of Genetic Screening

This audio clip discusses some of the advantages and disadvantages associated with genetic screening practices in humans.

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WWW WEB Activity

Simulation—Amniocentesis

Amniocentesis involves removing cells from the amniotic fluid, without damaging the fetus. Watch this animated simulation of amniocentesis to see how the cells are gathered and how they are used.

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INVESTIGATION 19.1 *Introduction*

Sex-Linked Traits

In this activity, you will cross *Drosophila* that carry genes for sex-linked traits, using virtual fruit fly software. To determine if a trait is sex-linked, you will perform two sets of crosses. In the first set of crosses, you will confirm that a trait is sex-linked using males and females with and without a trait. How will you set up the crosses to get the data you will need? In the second set of crosses, you will determine the phenotypic ratios in offspring of the F_1 generation and observe the frequency of one trait in the male and in the female offspring. What ratio would you expect for a sex-linked trait?

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	○ Synthesis
● Prediction	● Evidence	

To perform this investigation, turn to page 652.

SUMMARY *Chromosomes and Genetics*

- The chromosomal theory of inheritance:
  - Chromosomes carry genes, the units of heredity.
  - Each chromosome contains many different genes.
  - Paired chromosomes segregate during meiosis. Each sex cell or gamete has half the number of chromosomes found in a somatic cell.
  - Chromosomes assort independently during meiosis. This means that each gamete receives one member from each pair of chromosomes, and that each chromosome pair has no influence on the movement of any other chromosome pair.
- Females have two X chromosomes. Males have one X and one Y chromosome.
- Sex-linked traits are controlled by genes located on the sex chromosomes. A recessive trait located on the X chromosome is more likely to express itself in males than in females, since males need only one copy of the recessive allele while females need two.
- Female somatic cells can be identified by Barr bodies, which are actually dormant X chromosomes.

Section 19.1 Questions

1. Describe how the work of Walter S. Sutton and Theodor Boveri advanced our understanding of genetics.
2. How do sex cells differ from somatic cells?
3. Describe how Thomas Morgan's work with *Drosophila* advanced the study of genetics.
4. Identify two different sex-linked traits in humans.
5. What are Barr bodies?
6. A recessive sex-linked allele *(h)* located on the X chromosome increases blood-clotting time, causing hemophilia.
   (a) With the aid of a Punnett square, explain how a hemophilic offspring can be born to two normal parents.
   (b) Can any of the female offspring develop hemophilia? Explain.
7. In humans, the recessive allele that causes a form of red–green colour-blindness *(c)* is found on the X chromosome.
   (a) Identify the F_1 generation from a colour-blind father and a mother who is homozygous for colour vision.
   (b) Identify the F_1 generation from a father who has colour vision and a mother who is heterozygous for colour vision.
   (c) Use a Punnett square to identify parents that could produce a daughter who is colour-blind.

Gene Linkage and Crossover 19.2

It is often said that great science occurs when good questions are asked. Like Mendel, Morgan asked great questions when he observed a few unexpected gene combinations when he performed some dihybrid crosses with *Drosophila*. Morgan had found a number of obvious mutations in *Drosophila*. He had noted a number of genes in *Drosophila* that had different alleles that were easy to observe, which he used in many genetic experiments. When he carried out dihybrid crosses of *Drosophila*, Morgan observed that in some of the crosses, almost all the offspring had the same combination of traits as did the parents. Morgan's hypothesis to explain these observations, which he tested with further experiments, gave further support to the theory that the genes are located on chromosomes.

Morgan first crossed *Drosophila* homozygous for wild-type body-colour (*AA*) and straight wings (*BB*) with *Drosophila* homozygous for black body-colour (*aa*) and curved wings (*bb*). The resulting F_1 generation was therefore heterozygous for both traits (*AaBb*). When members of the F_1 generation mated among themselves, the F_2 generation showed far less variability than expected. Since this was a dihybrid cross, Morgan had predicted that the F_2 generation would have a 9:3:3:1 phenotypic ratio, as was observed in the work of Mendel. Instead, nearly all the individuals with wild-type body-colour had straight wings and nearly all those with black body-colour had curved wings.

Why did the observed ratios differ so much from the predicted ratio? From these observations, Morgan concluded that the two genes must not have undergone independent segregation. For this to be true, both genes would have to be located on the same chromosome. In other words, the genes for body colour and wing shape must be linked genes.

Figure 1 illustrates what would happen to the alleles in this cross during meiosis, if Morgan's hypothesis was correct and the genes for body colour and wing shape were linked genes.

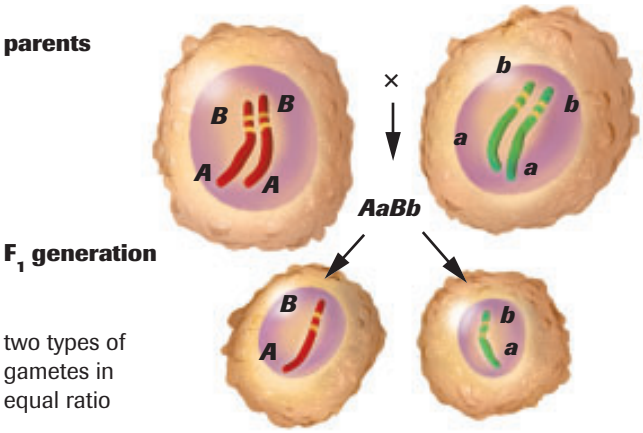


Figure 1
During meiosis, homologous chromosomes (represented as green and red chromosomes) move to opposite poles. One gamete carries the *AB* alleles and the other carries the *ab* alleles.

When two gametes from this cross unite, the new individual is heterozygous for both traits (*AaBb*). Remember that one parent carried the dominant alleles of the two linked genes (*A* is linked to *B*) and the other parent carried the recessive alleles (*a* is linked to *b*). Morgan, therefore, predicted that the F_2 generation would have a 3:1 phenotypic ratio (three flies with wild-type body-colour and straight wings to every one with black body-colour and curved wings), as shown in **Figure 2**, on the next page.

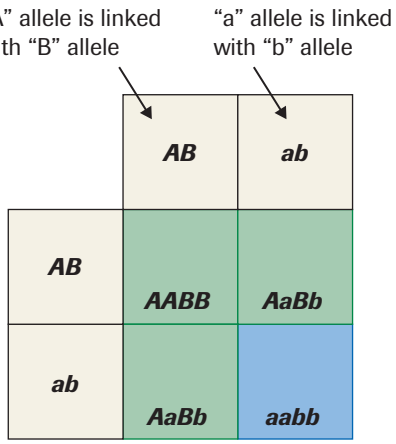


Figure 2
Punnett square analysis, assuming that all the gametes carry the same alleles as the parent. The expected phenotypic ratio is three wild-type body-colour, straight wings to one black body-colour, curved wings.

Morgan was able to find a number of linked genes. Some of these are shown in **Table 1**.

Table 1 Linked Genes Identified by Morgan's Research on *Drosophila*

Trait	Dominant/Recessive	Location
wingless *(wg)*	recessive lethal (all wingless offspring are born dead)	chromosome 2
curly wings *(Cy)*	dominant	chromosome 2
purple eyes *(pr)*	recessive nonlethal	chromosome 2
stubble bristles *(Sb)*	dominant	chromosome 3
ebony body *(e)*	recessive nonlethal	chromosome 3
miniature wings *(m)*	sex-linked recessive	chromosome 4
cut wings *(ct)*	sex-linked recessive	chromosome 4
white eyes *(w)*	sex-linked recessive	chromosome 4
vermillion eyes *(v)*	sex-linked recessive	chromosome 4

Crossing Over

Mendel had explained most of his observations by hypothesizing that the two genes were both on the same chromosome. By the Punnett square analysis shown in **Figure 2**, only two different phenotypes are predicted for these linked genes. This was not what Morgan observed. In a small number of flies from the dihybrid cross, the offspring had a different combination of traits than the parents. **Table 2** shows the numbers of the different phenotypes and their predicted genotypes. Where did the new allele combinations come from? Where did the new combinations of the two traits come from?

Table 2 Observed Progeny (F_2) of *AaBb* × *AaBb* F_1 Parents

Phenotype	Number	Possible genotype
wild-type body-colour, straight wings	290	*AABB* or *AaBb*
black body-colour, curved wings	92	*Aabb*
wild-type body-colour, curved wings	9	*AAbb* or *Aabb* indicated recombinations
black body-colour, straight wings	9	*AaBB* or *aaBb* indicated recombinations

Recall that chromosomes sometimes undergo crossing over during meiosis. During crossing over, a segment of DNA on one homologous chromosome is exchanged with the corresponding segment on the other homologous chromosome (**Figure 3**), recombining the set of genes on the chromosomes. Crossing over occurs in meiosis, during synapsis. Through crossing over, the gene combinations on a single chromosome can be altered as it is passed from generation to generation. In this cross, gametes with the gene combination *Ab* and *aB* would not occur without crossing over.

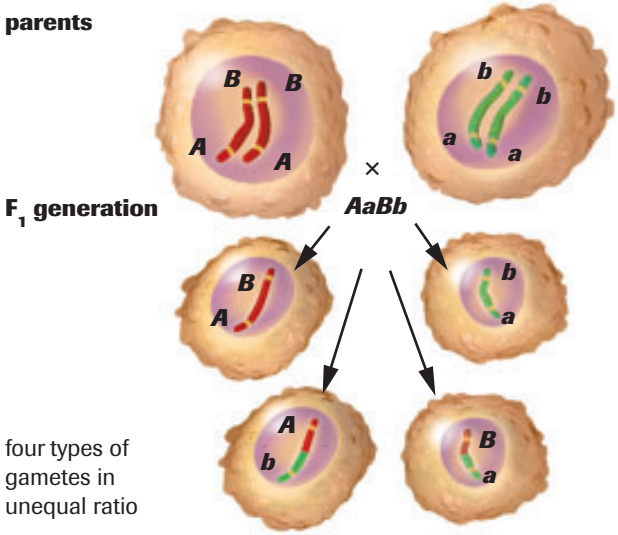


Figure 3
Consider the green chromosome to have been inherited from the father and the red from the mother. In the gametes, a chromosome that has undergone crossing has sections that are maternal (coming from the mother) and sections that are paternal (coming from the father). When the maternal and paternal homologous chromosomes carry different alleles, they may exchange alleles.

Mapping Chromosomes

As other traits in *Drosophila* were studied, it became clear that there were groups of linked genes. These **linkage groups** corresponded to different chromosomes. Furthermore, particular genes were always found at the same location (**locus**) on the chromosome. If this were not true, crossing over would not result in the exact exchange of alleles.

linkage group a group of linked genes on a chromosome

locus (plural, **loci**) a specific location along a chromosome where a particular gene is found

Morgan's experiments also showed that the frequency of crossovers between any two genes in a linkage group was always the same. The frequency of crossing over between any two genes can be stated as a percent:

$$\text{crossover percentage} = \frac{\text{number of recombinations}}{\text{total number of offspring}} \times 100\ \%$$

The crossover percentage in the offspring shown in **Table 2**, on the previous page, is

$$\text{crossover percentage} = \frac{18}{400} \times 100\ \%$$

$$= 4.5\ \%$$

The percentage of crossovers is related to the actual physical distance of the two genes on the chromosome. Genes located farther away from one another cross over at higher frequencies than genes located close together. Two genes with a crossover percentage of 1 % are much closer to one another than two genes with a crossover percentage of 12 %. Armed with this knowledge, geneticists were able to build a map of the chromosomes of *Drosophila* (**Figure 4**, next page).

When genes are in the correct order on a chromosome map, the map distances between the different genes is additive. This fact allows us to place genes in their proper order, based on the percentage crossover values between the different genes.

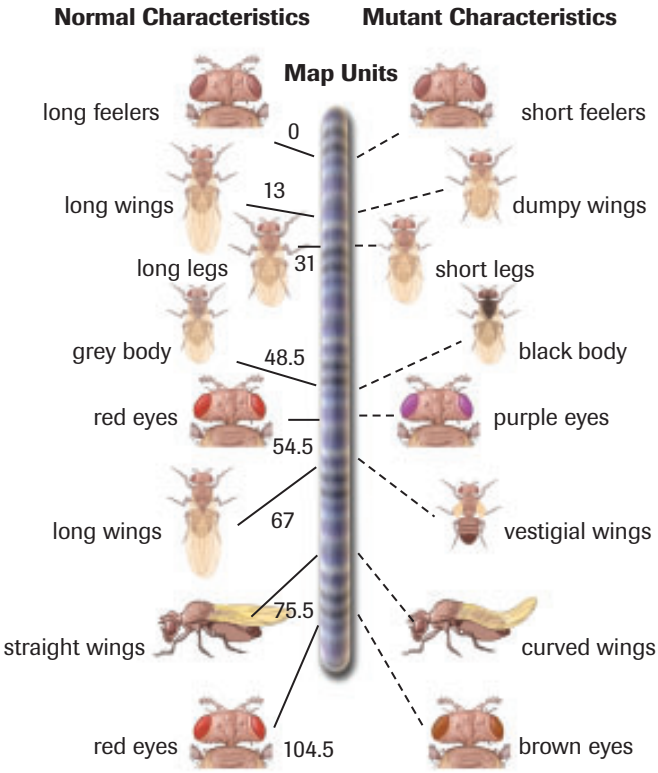


Figure 4
Gene mapping of chromosome 2 for *Drosophila melanogaster*. Note that many genes are located on one chromosome.

SAMPLE exercise 1

From crosses between different *Drosophila*, a geneticist finds that the crossover frequency between gene *A* and gene *B* is 12 %, the crossover frequency between gene *B* and gene *C* is 7 %, and between gene *A* and gene *C* is 5 %. What is the order and relative distances of these three genes on the chromosome?

Solution

If gene A were in the middle, then the sum of the distances between B and A and A and C must equal the distance between B and C. These distances are not equal, so A is not in the middle **(Figure 5)**.

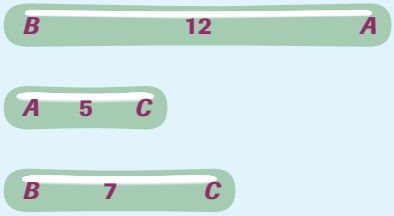
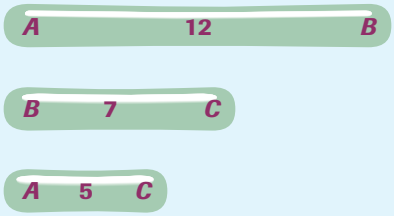


Figure 5

$BA + AC \neq BC$
$12 + 5 \neq 7$

Therefore, *A* is not in the middle.

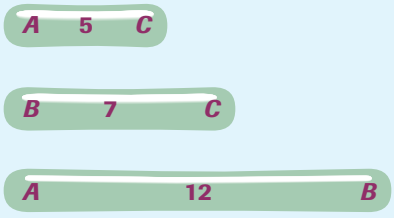
If gene B were in the middle, then the sum of the distances between A and B and between B and C must equal the distance between A and C. These distances are not equal, so B is not in the middle (**Figure 6**, next page).



$AB + BC \neq AC$
$12 + 7 \neq 5$

Therefore, *B* is not in the middle. **Figure 6**

If gene C were the middle gene, then the sum of the distances between A and C and C and B must equal the distance between A and B. These distances are equal. Therefore, C is in the middle **(Figure 7)**.



$AC + CB = AB$
$5 + 7 = 12$

Therefore, *C* is in the middle. **Figure 7**

Practice

1. A geneticist observes that the crossover frequency between gene A and gene B is 4 %, the crossover frequency between gene B and gene C is 14 %, and between gene A and gene C is 10 %. What is the order and relative distances of these three genes on the chromosome?

LAB EXERCISE 19.B

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	○ Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	● Evidence	

Mapping Chromosomes

A. H. Sturtevant, a student who worked with Thomas Morgan, hypothesized that

- genes are located in a linear series along a chromosome, much like beads on a string,
- genes that are closer together will be separated less frequently than those that are far apart,
- and that crossover frequencies can be used to construct gene maps.

Sturtevant's work with *Drosophila* helped establish techniques for chromosome maps.

Procedure

1. Examine the picture of a chromosome (**Figure 8**, next page). Crossing over takes place when breaks occur in the chromatids of homologous chromosomes during meiosis. The chromatids break and join with the chromatids of homologous chromosomes. This causes an exchange of alleles between chromosomes.

LAB EXERCISE 19.B *continued*

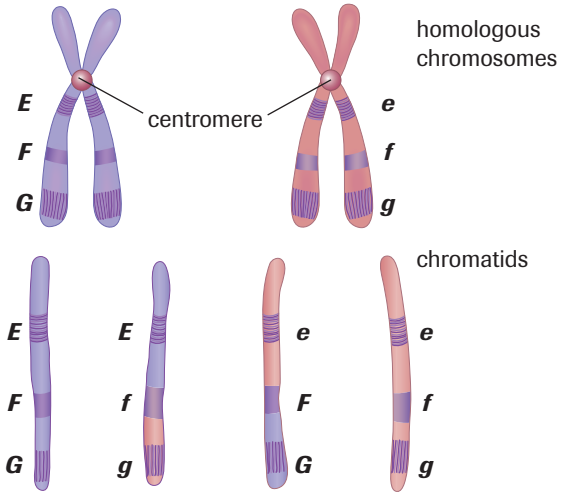


Figure 8
Crossing over

(a) Circle the areas of the chromatids that show crossing over.

(b) Using the diagram above, which genes appear farthest apart? (Choose from *EF*, *FG*, or *EG*.)

(c) Which alleles have been exchanged?

2. In 1913, Sturtevant used crossover frequencies of *Drosophila* to construct chromosome maps. To determine map distances, he arbitrarily assigned one recombination for every 100 fertilized eggs. For example, genes that had a crossover frequency of 15 % were said to be 15 units apart. Genes that had a 5 % recombination rate were much closer. These genes are 5 units apart.

(d) Using the data in **Table 3**, determine the distance between genes *E* and *F*.

Table 3

Cross	Offspring	Frequency (%)
EF × *ef*	*EF* + *ef* (from parent)	94
	Ef + *eF* (recombination)	6

(e) Would the distance between genes *e* and *f* be identical?

3. Use the data in **Table 4** to construct a complete gene map.

(f) What is the distance between genes *E* and *G*?

(g) What is the distance between genes *F* and *G*?

Table 4

Cross	Offspring	Frequency (%)
EF × *ef*	*EF* + *ef* (from parent)	94
	Ef + *eF* (recombination)	6
EG × *eg*	*EG* + *eg* (from parent)	90
	Eg + *eG* (recombination)	10
FG × *fg*	*FG* + *fg* (from parent)	96
	Fg + *fG* (recombination)	4

Analysis

(h) What mathematical evidence indicates that gene *F* must be found between genes *E* and *G*?

(i) Draw the gene map to scale. (Use 1 cm to represent 1 unit.)

(j) For a series of breeding experiments, a linkage group composed of genes *W*, *X*, *Y*, and *Z* was found to show the gene combinations in **Table 5**. (All recombinations are expressed per 100 fertilized eggs.)

Table 5

Genes	*W*	*X*	*Y*	*Z*
W	-	5	7	8
X	5	-	2	3
Y	7	2	-	1
Z	8	3	1	-

Construct a gene map. Show the relative positions of each of the genes along the chromosome and indicate distances in map units.

(k) For a series of breeding experiments, a linkage group composed of genes *A*, *B*, *C*, and *D* was found to show the gene combinations in **Table 6**. (All recombinations are expressed per 100 fertilized eggs.) Construct a gene map. Show the relative positions of each of the genes along the chromosome and indicate distances in map units.

Table 6

Genes	*A*	*B*	*C*	*D*
A	-	12	15	4
B	12	-	3	8
C	15	3	-	11
D	4	8	11	-

Using Marker Genes

Earlier in the chapter, you learned that genes located on the same chromosome are usually inherited together. **Marker genes** can be used to follow the inheritance of a linked trait. Marker genes give rise to an easily identifiable phenotype and are used to trace the inheritance of other genes that are difficult to identify. The marker gene must be located on the same chromosome and, ideally, at a very small distance from the gene being traced.

marker gene a gene that confers an easily identifiable phenotype and is used to trace the inheritance of other genes that are difficult to identify; it must be located on the same chromosome, and ideally, at a very small distance from the gene being followed

Dr. Ram Mehta, president of PBR Laboratories in Edmonton, uses gene markers to identify possible gene mutations in yeast. The yeast cells are treated with agents that might alter the genetic structure of the yeast, such as various chemicals, or environmental agents such as radiation. Since the chemical structure of DNA in human chromosomes and yeast chromosomes is the same, the yeast provides a model that helps scientists to predict how any given agent may affect human chromosomes.

Normally, yeast colonies are an off-white colour. This colour is determined by a dominant gene. Pink or red colonies indicate that a mutation in this normal, dominant gene has taken place (**Figure 9**). The red and pink colour is determined by one of two marker genes that are located along different sections of the chromosome. The marker genes are expressed only when the normal, dominant gene for colour has been inactivated by a mutation. Colonies will show both pink and red colour only when crossing over has occurred. Crossing over indicates that the agent being tested broke apart the yeast chromosome containing the marker genes. Mutation rates can be calculated from the frequency with which pink or red colonies appear.

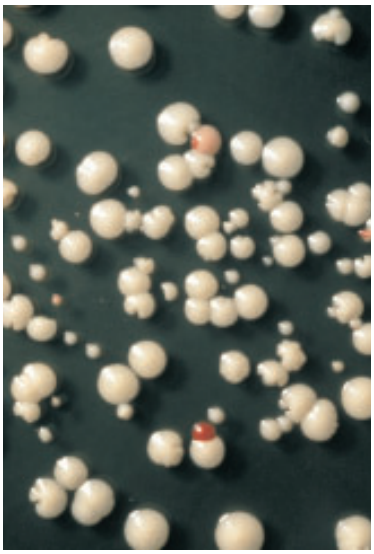


Figure 9
Mutated yeast colonies

SUMMARY Gene Linkage and Crossover

- Linked genes do not segregate independently because they are situated on the same chromosome. Linked genes can undergo recombination due to crossing over.
- Crossing over occurs more frequently between genes located relatively far apart than for those located relatively close together.
- Genetic linkage maps can be created by sorting genes according to the percentage crossover values.

Section 19.2 Questions

1. Why does gene linkage limit the variability of an organism?
2. Does crossing over increase or decrease the variability of an organism? Explain.
3. Create a chromosome map for each set of three genes from the given information.
   (a) The crossover frequency between gene A and gene B is 23 %, the crossover frequency between gene B and gene C is 11 %, and between gene A and gene C is 12 %.
   (b) The crossover frequency between gene X and gene Z is 8.5 %, the crossover frequency between gene Y and gene Z is 2.25 %, and between gene Y and gene X is 6.25 %.

19.3 DNA Is the Hereditary Material

Figure 1
DNA contains the information that ensures that pea plants produce seeds that grow into other pea plants.

continuity of life a succession of offspring that share structural similarities with those of their parents

The nucleus of every cell in your body contains deoxyribonucleic acid, or DNA. DNA is found in the cells of all organisms, from mushrooms to trees, from sponges to mammals. Scientists' fascination with DNA arises from the fact that it is the only molecule known that is capable of replicating itself. Sugar molecules, protein molecules, and fat molecules cannot build duplicates of themselves. DNA can duplicate itself, thereby permitting cell division.

Sometimes referred to as the language of life, the genetic code is contained in 46 separate chromosomes in your body. Characteristics such as your hair colour, skin colour, and nose length are all coded within the chemical messages of DNA. Packed within the DNA are all the instructions that make you unique. Unless you are an identical twin, your DNA code is distinctively one of a kind.

DNA contains instructions that ensure **continuity of life**, which we observe as similar structural traits between members of different generations. Pea plants produce seeds that grow into other pea plants because the DNA holds the chemical messages for the roots, stems, leaves, and seed pods of a pea (**Figure 1**). In a similar way, guinea pigs give birth to other guinea pigs, and humans procreate with other humans. However, you have learned that not all offspring are identical to their parents. The uniqueness of descendants can be explained by new combinations of genes and by mutations. In order to understand how genes affect the expression of an organism's traits, you will have to learn how DNA regulates the production of protein. Proteins are the structural components of cells. DNA, therefore, not only provides continuity of life, but also accounts for the diversity of life forms.

Finding the Material of Heredity

In 1869, twenty-five-year-old Swiss biochemist Friedrich Miescher extracted a viscous white substance from white blood cells deposited on the bandages of wounded soldiers. He named this slightly acidic, phosphorus and nitrogen-rich material nuclein because he found it within the nuclei of these cells. With further work, Miescher found that nuclein was comprised of both an acidic portion, which he called nucleic acid, and an alkaline portion. The alkaline portion was later determined to be protein. Several decades later, Miescher's single nucleic acid was shown to actually be two nucleic acids, one of which was renamed ribonucleic acid (RNA) and the other, deoxyribonucleic acid (DNA). Ongoing research gradually revealed the structure, function, and importance of the remarkable and complex DNA molecule and showed it to be the source of hereditary information. This knowledge in turn triggered revolutions in the biological sciences.

Early work aimed at finding the material of heredity focused on proteins as the most probable source. In 1943, Danish biologist Joachim Hammerling demonstrated that the nucleus was likely to be the region in which the hereditary material of the cell would be found. He was able to do this as a result of research involving the large single-celled green alga *Acetabularia*. This organism grows to an average length of 5 cm and has three distinct regions known as the cap, the stalk, and the foot.

Hammerling's experiments first involved cutting the cap off of some cells and the foot, which contains the nucleus, off of others. The cells whose caps were removed were able to regenerate new caps, but the cells whose feet had been removed were not able to regenerate new ones (**Figure 2**, next page). As a result, Hammerling hypothesized that the hereditary information was contained in the foot and, more specifically, the

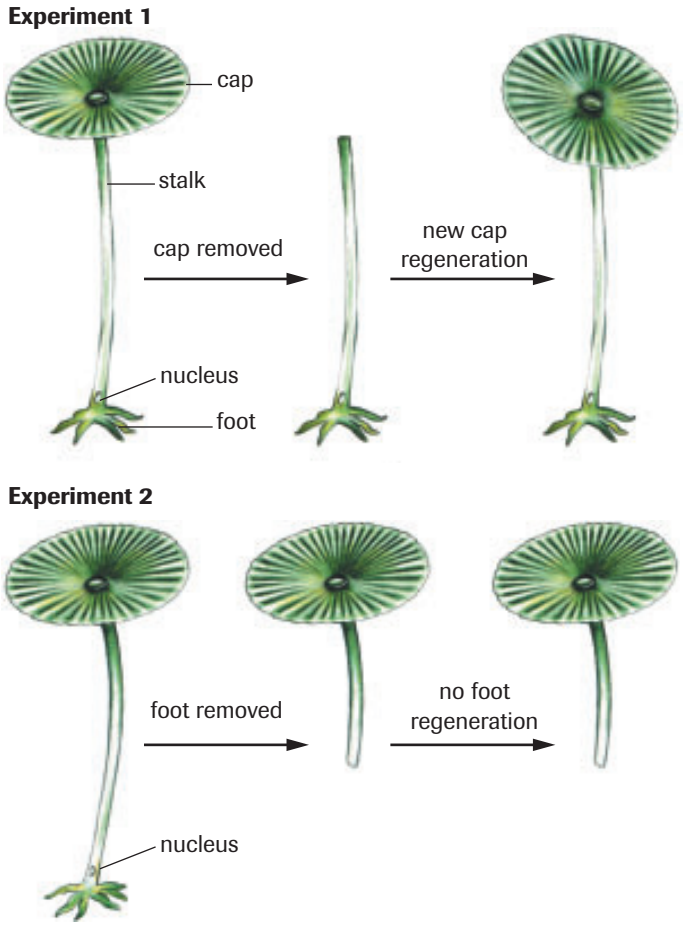


Figure 2
Hammerling's experiment strongly suggested that the hereditary material is located in the nucleus.

nucleus. To further test his hypothesis, he conducted additional experiments in which he transplanted stalks from a species of *Acetabularia* with a flowerlike cap onto the foot of another species with a disk-shaped cap. The caps that eventually developed on the transplanted stalks were all disk-shaped. Hammerling concluded that the instructions needed to build these new caps were very likely in the nucleus in the foot of the cell and not elsewhere.

Hammerling's results encouraged scientists to concentrate their search for the material of hereditary material on the nucleus and its contents. Proteins and DNA are present in the nucleus in large quantities, but DNA was initially thought to be too simple a material to account for the great variety seen in cells and cell processes, while proteins were already known to play a significant role in metabolic functions. However, work by British biologist Frederick Griffith on *Streptococcus pneumoniae*, in 1928, laid the foundation for later research. Canadian-born scientists Oswald Avery and Colin MacLeod, along with their American teammate Maclyn McCarty, built upon this work over a 14-year period culminating in 1944, and came to the conclusion that DNA was indeed the molecular material of heredity.

DID YOU KNOW?

One Man's Castle Is Another Man's Lab

Friedrich Miescher's discovery took place in the vaults of an old castle that had been converted to a laboratory. You can hear Miescher describe the process he used to isolate nuclein in an animation found by accessing the Nelson Web site.

www.science.nelson.com GO

DID YOU KNOW?

DNA's Homes

DNA does not just reside in the nucleus. A small amount of DNA is also found in chloroplasts and mitochondria. The size of the genome varies depending on the species. Plants tend to have a larger mitochondrial genome compared with that of animals.

LAB EXERCISE 19.C

Evidence of Hereditary Material

In the 1920s, Frederick Griffith, an English medical officer, started experimenting with *Streptococcus pneumoniae*. This bacterium, which causes pneumonia, exists in two forms. One form is surrounded by a polysaccharide coating called a capsule and is known as the S form because it forms smooth colonies on a culture dish. The second harmless form has no coating and is known as the R form because it forms rough colonies on a culture dish (**Figure 3**).

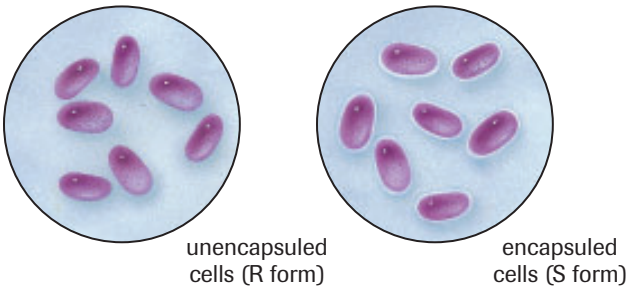


Figure 3
A representation of the two forms of *S. pneumoniae*

Report Checklist

○ Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	● Synthesis
○ Prediction	○ Evidence	

The following is an abbreviated summary of Griffith's procedures and results:

Procedure

1. Mouse A was injected with encapsulated cells (S form), while mouse B was injected with unencapsulated cells (R form).
2. Encapsulated (S-form) pneumococcal cells were heated, killed, and then injected into mouse C (**Figure 4**).
3. The heated encapsulated (S-form) cells were mixed with unencapsulated (R-form) cells. The mixture was grown on a special growth medium. Cells from the culture medium were injected into mouse D (**Figure 4**).

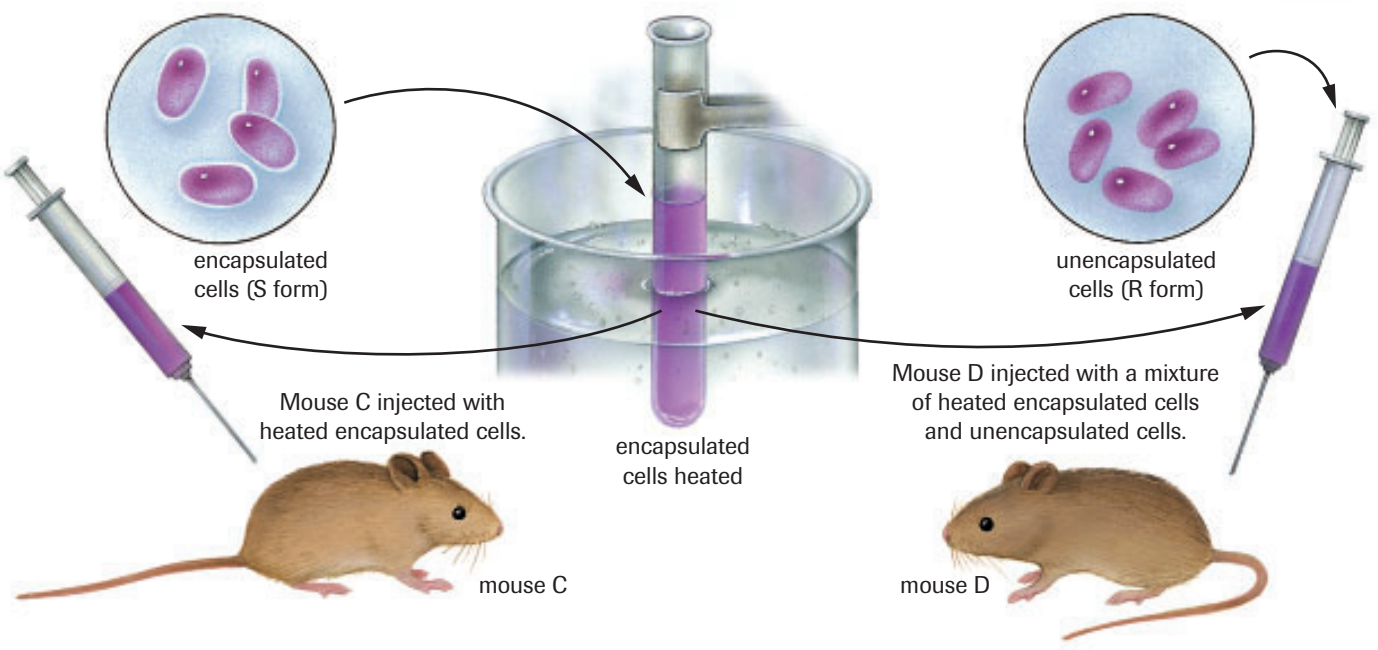


Figure 4
A visual outline of the procedure

LAB EXERCISE 19.C *continued*

Evidence

- Mouse A contracted pneumonia and died, while mouse B continued to live. Mouse B was sacrificed, and an autopsy was conducted on both mice. The autopsies revealed living S cells in mouse A's tissues and living R cells in mouse B's tissues.
- Mouse C continued to live. Mouse C was sacrificed and the autopsy revealed that no living S cells were found in the animal's tissues.
- Mouse D died. An autopsy indicated that the mouse had died of pneumonia; encapsulated (S-form) bacteria and unencapsulated (R-form) bacteria were isolated from the mouse.

Analysis and Evaluation

(a) What conclusions can you derive from the experimental results with mouse A and mouse B?

(b) Why might a scientist decide to repeat step 1 of this experimental procedure on other mice?

(c) What is the significance of the result with mouse C?

(d) Predict what would have happened to the mouse if the unencapsulated (R-form) cells had been heated and then injected. What would this step have represented in the experimental protocol?

(e) Would you have predicted that mouse D would die? Explain why or why not.

(f) A microscopic examination of the dead and live cell mixture (step 3) revealed cells with and without capsules. What influence did the heat-destroyed cells have on the unencapsulated cells?

(g) Griffith hypothesized that a chemical in the dead, heat-treated, encapsulated cells (step 3) must have altered the living unencapsulated cells and he dubbed this chemical phenomenon *transformation*. In 1944, Oswald Avery, Maclyn McCarty, and Colin MacLeod conducted experiments in test tubes with *Streptococcus pneumoniae* that led them to conclude that DNA is the *transforming principle*, as they called it, and not proteins, as was widely believed. In their experiments, what must have happened to the DNA when the cells divided?

Synthesis

(h) To discover the identity of the transforming principle, Avery and his associates ruptured heat-killed, encapsulated cells to release their contents. RNA, DNA, protein, and purified polysaccharide coats were isolated and were tested for transforming activity. Avery and his associates found that only R cells mixed with purified DNA isolated from dead S cells were transformed to S cells. When R cells were mixed with purified RNA, with the polysaccharide coat, or with protein extracted from dead S cells, only R cell colonies were isolated. Do these results support their hypothesis? Explain.

(i) Predict the experimental results of the following protocols. Support your prediction with a hypotheses.

- Polysaccharide-digesting enzymes are used to digest the encapsulated polysaccharide coat of the heated S form of the bacteria. The treated bacteria are then placed with unencapsulated pneumonia cells, which are then injected into a mouse.
- Heated encapsulated bacteria are treated with DNAase, a DNA-digesting enzyme. The treated bacteria are then mixed with unencapsulated pneumonia cells, which are injected into a mouse.
- All proteins are extracted from the heated encapsulated bacteria. The treated bacteria are then mixed with unencapsulated pneumonia cells, which are injected into a mouse.

(j) Based on the information provided, suggest improvements to the experimental protocols.

Figure 5
Dr. Oswald Avery

Figure 6
Dr. Colin MacLeod

bacteriophage a virus that infects bacteria

WEB Activity

Canadian Achievers—Avery and MacLeod

Canadian-born scientists Dr. Oswald Avery and Dr. Colin MacLeod spent their early years as scientists in Nova Scotia, where they were born. They met in New York, where, together with American scientist Maclyn McCarty, they painstakingly isolated components of pneumococci (*Streptococcus pneumoniae*) for over a decade before identifying DNA as the transforming principle. You can find more information on this classic experiment in an animation by accessing the Nelson science Web site.

www.science.nelson.com GO

Confirming the Chemical of Heredity

Frederick Griffith's work in the 1920s began because he was trying to develop a vaccine against pneumonia caused by *Streptococcus pneumoniae*. However, his unexpected experimental observations, followed by the work of Avery, McCarty, and MacLeod, led scientists to begin questioning the initial assumption within the scientific community that the material of heredity was protein. What was now needed was experimental evidence that would clearly and conclusively indicate that DNA was indeed the material of heredity. This evidence was to come some six years after the work of Avery's team as the result of an innovative experiment.

Alfred D. Hershey and Martha Chase

It was not until 1952 that DNA was accepted as the hereditary material. That year, American scientists Alfred Hershey and Martha Chase conducted experiments using a virus (**bacteriophage** T2) that infects a bacterial host (**Figure 7**). Bacteriophages (commonly called phages) consist of two components: DNA and a protein coat. A bacteriophage infects a bacterial cell by attaching to the outer surface of the cell and injecting its hereditary information into it. This leads to the production of thousands of new viruses, which then burst out of the cell, resulting in its death. The results of Hershey and Chase's experiments showed that only the DNA from the bacteriophage, and not the protein coat, enters the bacteria to direct the synthesis of new viral DNA and new viral protein coats.

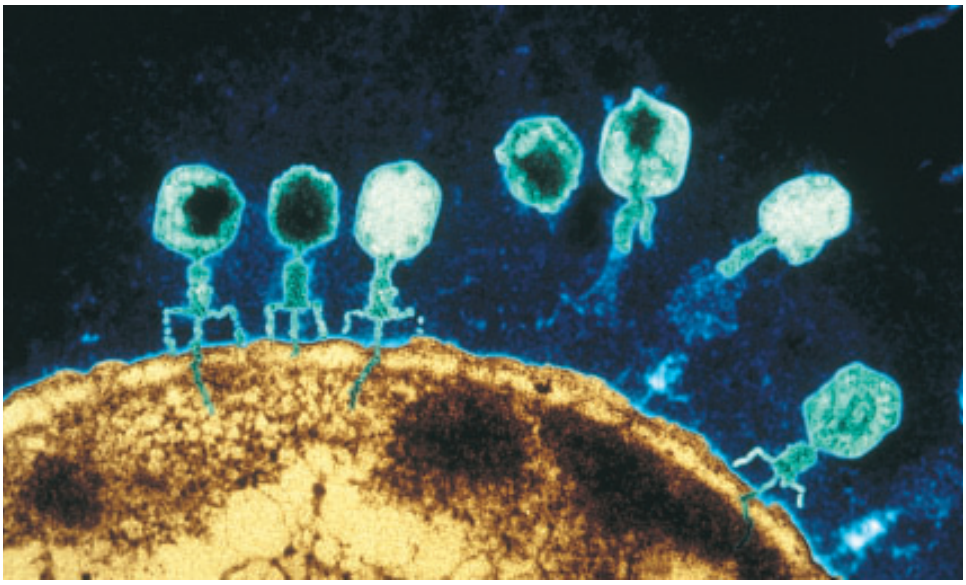


Figure 7
Micrograph of a bacteriophage injecting its DNA into a bacterial cell

Proteins contain sulfur but no phosphorus, whereas DNA contains phosphorus but no sulfur. Therefore, to track the location of DNA and proteins, Hershey and Chase tagged the viral proteins with an **isotope** of sulfur, ^{35}S, and the viral DNA with an isotope of phosphorus, ^{32}P. ^{35}S and ^{32}P are **radioisotopes** of sulfur and phosphorus, respectively. They are easy to track in an experiment because radioisotopes are unstable and the radiation that they emit as they decay can be measured.

isotope one of two or more atoms of the same element containing the same number of protons but a different number of neutrons

radioisotope an unstable isotope that decays spontaneously by emitting radiation

Each type of tagged bacteriophage was allowed to infect a separate batch of bacterial host cells and to multiply. The bacterial cells were put into a blender to remove the protein coats of the viruses from the surfaces of the bacteria. The mixtures were then subjected to centrifugation to isolate the individual components (bacteria as a pellet and viral particles in the liquid). The bacterial cells that were exposed to viruses containing radioactively labelled DNA contained ^{32}P. The bacterial cells that were exposed to viruses whose protein coats were radioactively tagged with ^{35}S did not contain any radioactivity; instead, the radioactive ^{35}S was found in the culture medium (**Figure 8**). These experiments illustrate that phosphorus-rich DNA was injected into the bacterial cells. In addition, Hershey and Chase found that the bacteriophages in both experiments reproduced and destroyed the bacterial cells that they had infected. This observation further supported the claim that DNA entering the host bacterial cell carries all the genetic information. Hershey and Chase's experiments ended the debate. DNA was accepted as the hereditary material.

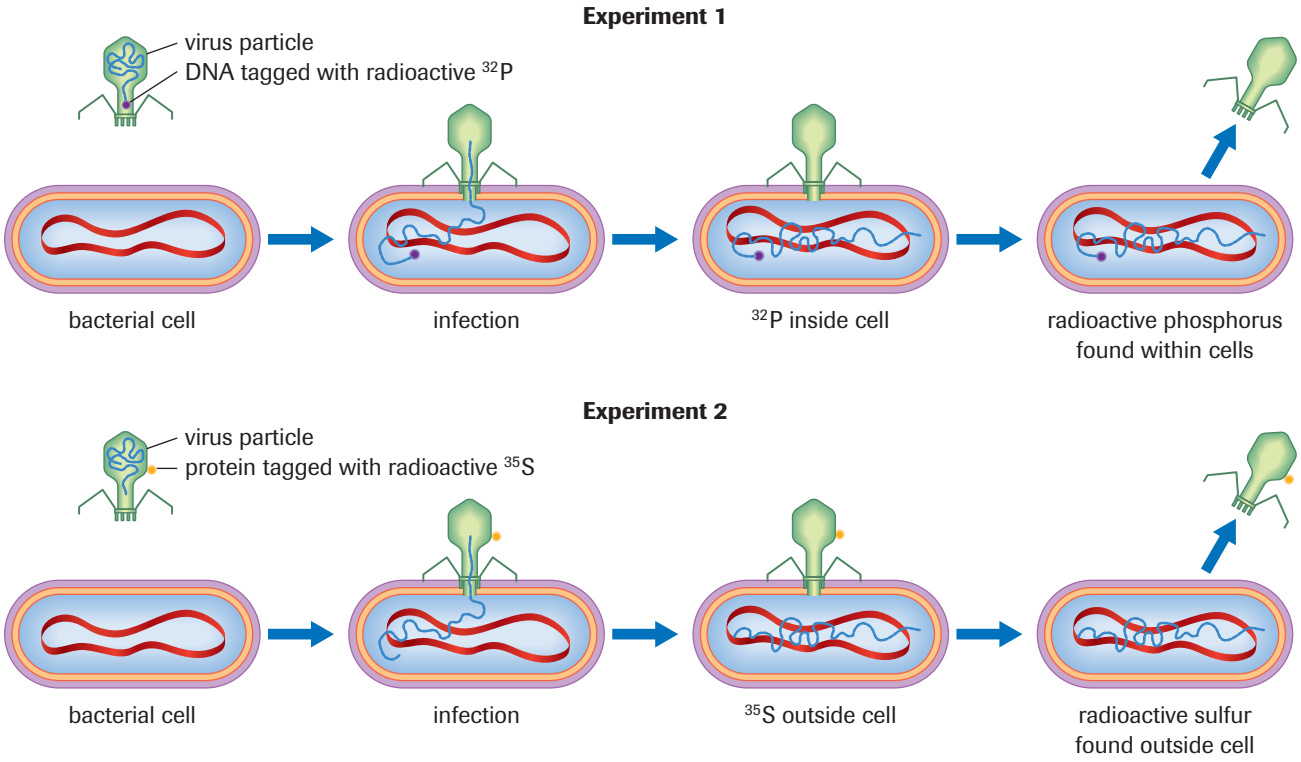


Figure 8
Hershey and Chase's experiment conclusively showed that DNA was the hereditary material.

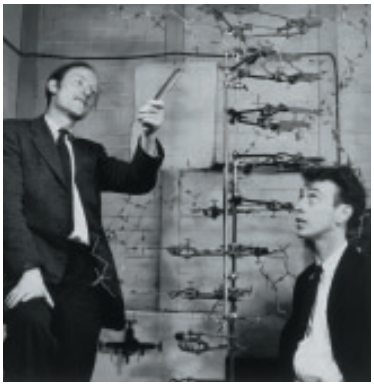


Figure 9
Francis Crick and James Watson were awarded the Nobel Prize for Physiology or Medicine in 1962 for deducing the structure of DNA.

The Race to Reveal the Structure

When scientists confirmed that DNA was the material of heredity, their focus shifted to understanding how it works. Part of that understanding would come from knowing its structure since, as in other subjects, structure in biology provides many clues about function. In the race to be the first to discover the structure of DNA, scientists around the world employed emerging technologies to help them gain new insights into this mysterious "molecule of life." In the end, the honour would go James Watson and Francis Crick (**Figure 9**).

James Watson was considered a child prodigy when he entered the University of Chicago at the age of 15. He began studying ornithology, but eventually turned his attention to genetics and molecular biology. In 1951, he began studies at England's Cambridge University, where he met Francis Crick, a physicist who had served with the British army during World War II. Each would bring to bear his experience from a different area of science to interpret and synthesize the experimental data that were rapidly mounting.

One source of important data came from the Cambridge laboratory of Maurice Wilkins, where researcher Rosalind Franklin used a technique called X-ray diffraction to help determine the structure of the DNA molecule. Another source of data involved the comparison of the chemical structure of DNA molecules in different organisms. By this time it had long been known that DNA is comprised of chains of molecules called **nucleotides**. The nucleotides consist of a 5-carbon cyclic ring structure called a **deoxyribose sugar** (**Figure 10**) having one of four **nitrogenous bases** attached to its 1′ carbon and a **phosphate group** attached to it 5′ carbon (**Figure 11**). The carbons in the sugar are identified by the numbers one to five and a prime (′) symbol to distinguish them from the carbons in the nitrogenous base. The four nitrogenous bases are adenine (A), guanine (G), thymine (T), and cytosine (C). Adenine and guanine are double-ringed structures classed as purines, while thymine and cytosine are single-ringed structures classed as pyrimidines. The only difference in the nucleotides is in their bases.

nucleotide a molecule having a five-carbon sugar with a nitrogenous base attached to its 1′ carbon and a phosphate group attached to its 5′ carbon

deoxyribose sugar a sugar molecule containing five carbons that has lost the –OH (hydroxyl group) on its 2′ position

nitrogenous base an alkaline, cyclic molecule containing nitrogen

phosphate group a group of four oxygen atoms surrounding a central phosphorus atom found in the backbone of DNA

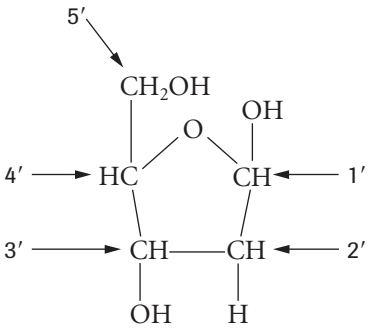


Figure 10
A deoxyribose sugar with numbered carbons

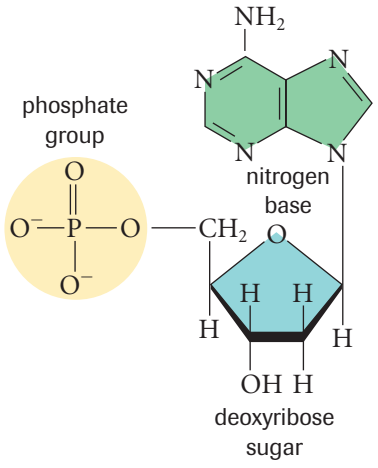


Figure 11
A DNA nucleotide is comprised of a deoxyribose sugar, a nitrogenous base, which in this case is adenine, and a phosphate group.

Biochemist Erwin Chargaff's evidence was crucial to helping Watson and Crick construct an accurate model of DNA. His observations determined that, for the DNA of any given species, the amount of adenine was always equal to the amount of thymine and the amount of guanine was always equal to the amount of cytosine. This relationship between the bases was consistent across all the species that he investigated. Although one species might have a different amount of adenine compared to another species, for example, the amount of thymine in each species was always equal to the amount of adenine.

Just as crucial was the X-ray photograph taken by Rosalind Franklin, which indicated that DNA was a helix that was likely double-stranded, that the distance between the strands was constant, and that the helix completed a full turn once every ten base pairs (**Figure 12**). Given this new data, Watson and Crick were able construct a three-dimensional scale model of DNA that portrayed the relationship between the bases as well as all of the nucleotide chemical bond angles and spacing of atoms consistent with the observations of other researchers. They presented their model to the scientific community in 1953, and in 1962 were awarded the Nobel Prize along with Maurice Wilkins. Because she had died prior to 1962 and the Nobel Prize is awarded only to living recipients, Rosalind Franklin was not included despite the acknowledgement of the significant importance of her photograph to the model proposed by Watson and Crick.

The Watson and Crick model of DNA structure is essentially the same one used by scientists today. Scientists already knew that molecules of DNA were made up of sugars (deoxyribose), phosphate, and four different nitrogen bases: adenine, guanine, cytosine, and thymine. What scientists did not know was the way in which these bases were arranged.

Learning Tip

Chargaff's Rules

The proportion of A always equals that of T (A = T).
The proportion of G always equals that of C (G = C).
A + G = T + C

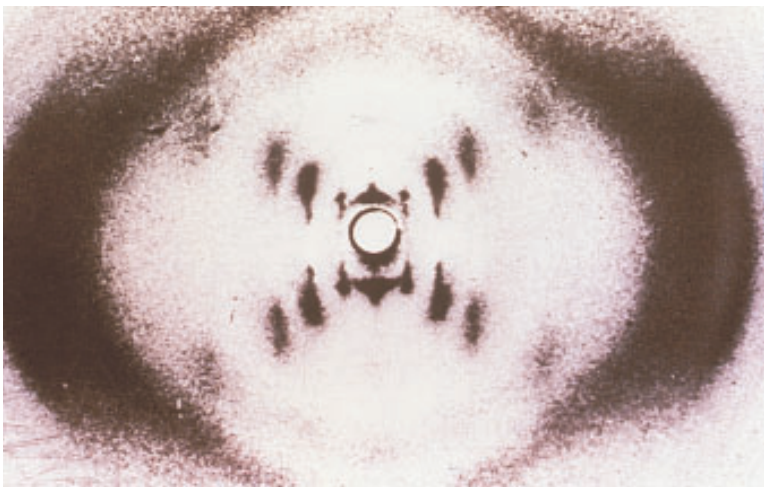


Figure 12
Rosalind Franklin's X-ray diffraction pattern of DNA revealed that it had a helical structure.

WEB Activity

Simulation—Elementary, My Dear Crick

Erwin Chargaff visited Watson and Crick in Cambridge in 1952. Crick's lack of knowledge with respect to nitrogenous bases did not impress Chargaff. By the following year, Watson and Crick had constructed their model of DNA. Enjoy Watson and Crick's deductive process in an animation found by accessing the Nelson Web site.

www.science.nelson.com GO

Figure 13
Rosalind Franklin's X-ray crystallography was crucial to the determination of the structure of DNA.

Politics and Science

Watson and Crick might not have been credited as the co-discoverers of DNA were it not for politics. The X-ray diffraction technique developed in England had been used by Maurice Wilkins and Rosalind Franklin (**Figure 13**) to view the DNA molecule. At that time, the American scientist Linus Pauling, a leading investigator in the field, was refused a visa to England to study the X-ray photographs. Pauling, along with others, had been identified by then U.S. Senator Joseph McCarthy as a communist sympathizer for his support of the anti-nuclear movement. Many scientists today believe that the United States passport office may have unknowingly determined the winners in the race for the discovery of the double-helix model of DNA.

The McCarthy era of the early 1950s is considered by many historians as a time of paranoia and repression. Many creative people had their careers stifled or destroyed because of their perceived association with communism. In most cases the charges were unfounded. It is perhaps ironic that, in 1962, Linus Pauling was awarded a Nobel Prize, this time for his dedication to world peace.

INVESTIGATION 19.2 *Introduction*

Isolation and Quantification of DNA

In this activity, you will extract DNA from both beef liver and onion cells in Parts 1 and 2. If your school has the necessary reagents and equipment, you will then have the option of testing for the presence of DNA in Part 3 and of determining its concentration using a spectrophotometer in Part 4. You will need to gather evidence and analyze and evaluate the results that you observe, and to then explain those results in writing. Heed all cautions and wear safety equipment as instructed.

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	● Evidence	

To perform this investigation, turn to page 653.

EXPLORE an issue

Competition and Collaboration Advance Science

Scientists have been described as intelligent, ambitious, and sometimes competitive. Yet, for science to progress, many individuals must work together in a collaborative, communicative atmosphere. Current science demands two conflicting ideologies: competition and collaboration. A fine balance is not necessarily struck between the two. Other factors that come into play are economics, politics, market demand, profit, and patriotism in times of war.

Statement
Competition is the key driving force of science, followed by collaboration.

- Form groups to research this issue. Prepare a position paper in point form that supports or disputes this statement, using a specific example. Some scientists and case studies that may be used include Robert Oppenheimer's and Phillip Morrison's role in the Manhattan Project; the perception of Linus Pauling as a communist and the denial of a visa for him to visit Watson and Crick in Cambridge; Craig Venter and Eric Lander leading opposing research teams in the Human Genome Project; and Fritz Haber's role in the production of deadly gases during World War I.
- Search for information in periodicals, on CD-ROMS, and on the Internet.

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- As a group, present your supported view in a class discussion.

Issue Checklist

○ Issue	○ Design	● Analysis
● Resolution	● Evidence	● Evaluation

SUMMARY DNA is the Hereditary Material

Year	Scientist	Experimental results
late 1860s	Friedrich Miescher	• isolated nonprotein substance from nucleus of cells; named this substance nuclein
1928	Frederick Griffith	• experimented using mice and two different strains of pneumococcus bacteria (virulent and nonvirulent); observed that when heat-treated virulent pneumococcus was mixed with nonvirulent pneumococcus and was injected into healthy mice, death resulted • discovered the process of transformation
1943	Joachim Hammerling	• experimented using green alga *Acetabularia;* observed that regeneration of new appendages was driven by the nucleus-containing "foot" of the alga • hypothesized that hereditary information is stored in the nucleus
1944	Oswald Avery, Maclyn McCarty, and Colin MacLeod	• demonstrated that DNA was the transforming principle of pneumococcus bacteria
1949	Erwin Chargaff	• discovered that in the DNA of numerous organisms the amount of adenine is equal to the amount of thymine, and the amount of guanine is equal to that of cytosine
1952	Alfred Hershey and Martha Chase	• used radioactively labelled viruses, infected bacterial cells; observed that the infected bacterial cells contained radioactivity originating from DNA of the virus, suggesting that DNA is hereditary material
1953	Rosalind Franklin	• produced an X-ray diffraction pattern of DNA that suggested it was in the shape of a double helix
1953	James Watson and Francis Crick	• deduced the structure of DNA using information from the work of Chargaff, Franklin, and Maurice Wilkins

Section 19.3 Questions

1. Describe how the experiments of Joachim Hammerling; Frederick Griffith; Oswald Avery, Maclyn McCarty, and Colin MacLeod; and Alfred Hershey and Martha Chase strengthened the hypothesis that DNA is the hereditary material.
2. Explain why Hammerling's experiment cannot be used as conclusive scientific evidence that DNA is the hereditary material.
3. Hammerling chose *Acetabularia* as a model organism for his experiment. Identify some of the characteristics of this green alga that rendered it an ideal organism. Scientists use model organisms in many of their experiments. Identify social, economic, and physical characteristics that would make an organism highly suitable for experimental research. Explain why humans do not make ideal research subjects.
4. Explain why it is important to study both the historic experiments that revealed genetic principles and the principles themselves. Support your reasons, using examples.
5. It can be argued that the repetition of experiments is a waste of time, money, and other valuable resources. Provide arguments that support and dispute this statement. Use examples from the experiments of Griffith and of Avery, McCarty, and MacLeod to strengthen your arguments.

INVESTIGATION 19.1

Sex-Linked Traits

In this activity, you will cross *Drosophila* (**Figure 1**) that carry genes for sex-linked traits using virtual fruit fly software. To determine if a trait is sex-linked, you will perform two sets of crosses: A and B.

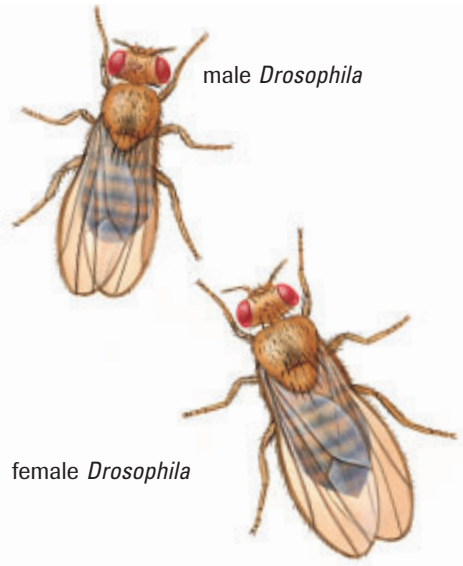

Figure 1
Drosophila males are smaller and have a rounded abdomen while the larger females have a pointed abdomen.

Familiarize yourself with the software before starting this activity. Start with the tutorial. Note that the labelling of traits in the software is different from the conventions used in this textbook. Be sure you understand what each label in the software correlates to in the textbook.

For cross A, these conditions must be met if the trait is sex-linked:

- In the F_1 generation, female offspring inherit the trait of the male parent and male offspring inherit the trait of the female parent.
- In the F_2 generation, there is a 1:1 phenotypic ratio for the traits in both males and females.

For cross B, you will confirm that the trait is sex-linked. You will cross parents with traits that are opposite to the traits of the parents in cross A. By examining the phenotypic ratios in offspring of the F_1 generation, you can observe the greater frequency of one trait in either the male or the female offspring.

Report Checklist

● Purpose	○ Design	● Analysis
○ Problem	○ Materials	● Evaluation
● Hypothesis	○ Procedure	○ Synthesis
● Prediction	● Evidence	

Problems

If white eye colour in *Drosophila* is a sex-linked recessive trait, what are the phenotypic ratios of the F_1 generation when a homozygous red-eyed female and a white-eyed male are crossed?

What other traits are sex-linked in *Drosophila*? Are they recessive or dominant?

Materials

virtual fruit fly simulation software
computers

Procedure

1. Log onto the software. Remember that each parent is homozygous for the trait chosen.
2. Select 1000 offspring.
3. For crosses A and B, follow these algorithms:
   A: P: white-eyed female × red-eyed male
   F_1: red-eyed female × red-eyed male
   F_2: red-eyed female × white-eyed male
   B: P: homozygous red-eyed female × white-eyed male
   F_1: red-eyed female × red-eyed male
4. For cross A, create a Punnett square to show the expected phenotypic ratio of offspring in each generation. Also, be sure to indicate the genotype of each phenotype.
5. After cross A, count the flies and the number of offspring (out of 1000) of each sex and with each trait. Record the information in a table beside the corresponding Punnett square.
6. When you have finished cross A, create a new parental generation.
7. Carry out cross B. Follow steps 4 to 6.
8. Determine if other traits are sex-linked. Follow the same procedure as in step 3, using new traits. Indicate which traits you are examining.

INVESTIGATION 19.1 *continued*

Analysis

(a) In one or two paragraphs, describe the results of crosses A and B. Is white eye colour in *Drosophila* sex-linked? If so, which sex does this trait appear in more frequently? Explain.

(b) In one or two paragraphs, describe the results with the other traits you examined. Is the trait sex-linked? If so, which sex does this trait appear in more frequently? Is the trait recessive or dominant? Explain.

Evaluation

(c) List and briefly explain any technical difficulties you had using the software.

(d) What improvements would you suggest to enhance the usefulness of the software?

(e) What are the advantages of using software to carry out this investigation compared to conducting it with actual *Drosophila*?

INVESTIGATION 19.2

Isolation and Quantification of DNA

In Parts 1 and 2 of this investigation, you will isolate DNA from onion cells and beef liver. Part 3 verifies the presence of DNA in your extraction using a biological analysis and Part 4 quantifies the amount of DNA using spectrophotometry. Parts 3 and 4 are optional depending on whether your school has the necessary reagents.

Report Checklist

● Purpose	● Design	● Analysis
○ Problem	○ Materials	● Evaluation
○ Hypothesis	○ Procedure	○ Synthesis
○ Prediction	● Evidence	

Problem

How much DNA can be extracted from plant and animal cells using simple laboratory methods?

Materials

safety goggles
rubber gloves
fresh beef liver
scissors
mortar and pestle
0.9 % (w/v) solution of sodium chloride (NaCl)
three 10 mL graduated cylinders
sand (very fine, washed)
cheesecloth
two 50 mL beakers, or two large test tubes
10 % (w/v) solution of sodium dodecyl sulfate (SDS)
95 % ethanol (chilled)
50 mL graduated cylinder
glass rod
four medium test tubes
4 % (w/v) solution of sodium chloride (NaCl)
onion
blender (optional)
hot-water bath
boiling chips
ice-water bath
meat tenderizer solution (3 g/50 mL of solution)
diphenylamine solution
25 mL graduated cylinder
Pasteur pipette, or plastic graduated eyedropper
distilled water
DNA standard solution
test-tube rack
spectrophotometer
cuvette
facial tissue

Procedure

DNA extraction is the first step in many biotechnological procedures. Cell walls and cell membranes must be disrupted to isolate DNA. In addition, lipids, proteins, and sugars must be separated from nucleic acid. In the following procedure, heat, detergents, salts, and cleaving enzymes are used to minimize contamination from nonnucleic acid molecules and to maximize purification.

Part 1: Extraction of DNA from Beef Liver

The ethanol solution is toxic and flammable. Keep it away from all sources of heat.

1. Obtain a 10 g to 15 g sample of beef liver and place it in the mortar.
2. Using scissors, cut the liver into small pieces.

INVESTIGATION 19.2 *continued*

3. Add 10 mL of 0.9 % NaCl solution to the diced liver. Use a 10 mL graduated cylinder to measure out the NaCl. Add a pinch of sand into the mixture to act as an abrasive, and grind the tissue thoroughly for approximately 5 min.
4. Strain the liver cell suspension through several layers of cheesecloth to eliminate any unpulverized liver. Collect the filtrate into a 50 mL beaker.
5. Add 3 mL of 10 % SDS solution. If a centrifuge is available, spin the suspension, and remove and save the supernatant. If a centrifuge is not available, mix the suspension thoroughly for 30 s and proceed to step 6.
6. Gently layer twice the volume (approximately 25 mL) of cold 95 % ethanol on the supernatant as that of the total volume of the cell suspension–SDS mixture. Use a 50 mL graduated cylinder to measure out the ethanol.
7. Using the glass rod, stir gently and slowly. A white, mucuslike substance will appear at the interface between the solutions. This substance is the DNA–nucleoprotein complex. After the complex has formed, twirl the stirring rod slowly and collect it onto the rod. Record your observations.
8. Place the isolated DNA–nucleoprotein complex into a test tube containing 3 mL of 4 % NaCl solution for later use. Use a 10 mL graduated cylinder to measure the 4 % NaCl solution. Pour the waste alcohol into the waste alcohol container designated by your teacher.

Part 2: Extraction of DNA from Onion

Onion is used because of its low starch content, which allows for a higher purity DNA extraction.

9. Repeat steps 1 to 5 using finely chopped onion. Instead of hand chopping the onion, a blender could be substituted, which gives optimum results.
10. Stir the mixture and let it sit for 15 min in a 60 °C water bath containing boiling chips. (Any longer and the DNA starts to break down.)
11. Cool the mixture in an ice-water bath for 5 min, stirring frequently.
12. Add half the volume of meat tenderizer solution as is present in your filtrate and swirl to mix.
13. Repeat steps 6 to 8.

Part 3: Testing for the Presence of DNA

The presence of DNA may be detected qualitatively with the reagent diphenylamine. Diphenylamine reacts with the purine nucleotides in DNA, producing a characteristic blue colour.

Diphenylamine solution contains glacial acetic acid. Be very careful not to spill any of the solution on yourself or on any surface. Inform your teacher immediately if any spills occur. Wear safety goggles and rubber gloves when handling this solution.

14. Stir the DNA from the onion and beef liver with their respective glass rods to resuspend them into the 4 % NaCl solution.
15. Dispense 15 mL of diphenylamine solution into a 25 mL graduated cylinder. The teacher will direct you to the stock diphenylamine solution, which will have been set up in a burette.
16. Transfer 5 mL of the solution to a 10 mL graduated cylinder with a Pasteur pipette or with a plastic graduated eyedropper.
17. Add 5 mL of diphenylamine solution to the DNA suspension obtained from the onion and from the beef liver.
18. Repeat step 16 and add 5 mL of diphenylamine solution to a test tube containing 3 mL of distilled water (the blank).
19. Repeat step 16 and add 5 mL of diphenylamine solution to a test tube containing 3 mL of DNA standard (the standard).
20. Place all of the test tubes in a boiling water bath (containing boiling chips) for 10 min and record the colour changes. Record your observations.
21. Remove the test tubes from the hot-water bath and place into a test-tube rack. Allow the tubes to cool before proceeding.

INVESTIGATION 19.2 *continued*

Part 4: Quantitative Determination of DNA Concentration Using Spectrophotometry

The principle underlying a spectrophotometric method of analysis involves the interaction of electromagnetic (EM) radiation (light) with matter. When EM radiation strikes an atom, energy in the form of light is absorbed. The remainder of the energy passes through the sample and can be detected. The more molecules that are present, the more energy will be absorbed, resulting in a higher absorbance reading. Since the relationship is direct, we can determine the concentration of an unknown by comparing it with a known. In this case, the unknown is the concentration of DNA in your samples and the known is the DNA standard.

22. Set the spectrophotometer to a wavelength of 600 nm. (See the video *Spec 20* on the Nelson Web site.)

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23. Fill a dry cuvette with the solution that consists of the distilled water and the diphenylamine. This will serve as a blank.
24. Wipe off any fingerprints from the outside of the cuvette by holding the cuvette at the very top and using a facial tissue. Place the blank into the spectrophotometer and set the absorbance to 0.00. (See the video *Spec 20* on the Nelson Web site.)

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25. Pour the blank solution back into its original test tube and place it in a test-tube rack.
26. Rinse the cuvette with a tiny amount of standard DNA solution (DNA standard and diphenylamine from step 19). Wipe off any fingerprints in the manner described in step 24.
27. Place the DNA standard solution into the spectrophotometer, then record the absorbance. (See the video *Spec 20* on the Nelson Web site.)

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28. Pour the DNA standard solution into its original test tube and save in case of error.
29. Repeat steps 26 to 28 with the beef liver extract solution and with the onion extract solution.

Analysis

(a) Propose reasons that the onion cells required heating and the liver cells did not.

(b) DNA was spooled out using a glass rod. How do you account for the "stickiness" of DNA to glass?

(c) Describe the DNA you extracted. If DNA is a rigid structure, why do the DNA strands appear flexible? What features of DNA's structure account for its stiffness? If DNA is rigid, how does it coil tightly into a small space?

(d) Comment on the purity of the DNA extracted.

(e) Compare the amount of DNA extracted from the onion versus that from the liver. Which source of DNA provided more of the molecule? Account for this observation, given your knowledge of cell structure and given differences in the procedure.

(f) What was the purpose of the standard DNA solution? What was the purpose of the blank?

(g) Did the spectrophotometric results correlate with the qualitative observations obtained from the diphenylamine test? Comment.

(h) Calculate the amount of DNA extracted from each source using your standard as a guide.

(i) The liver and onion were chopped very finely. Provide reasoning for this step. If the step was omitted, what effect would this omission have on the results?

(j) SDS is a detergent. Describe how detergents work and explain the role of SDS in the protocol.

(k) How does NaCl contribute to maximum DNA extraction? (Hint: Think about DNA's chemical constituents.) Keep in mind that NaCl is a salt that ionizes in solution.

(l) What is the purpose of adding cold ethanol to each extraction? How does this phenomenon work?

(m) In the extraction of DNA from onion, you added a meat tenderizer solution. The meat tenderizer solution contains an enzyme called papain. What role does papain play in the extraction?

(n) Identify three properties of DNA that are demonstrated by this investigation.

Evaluation

(o) Suggest possible sources of error in this procedure and describe their effect on the results.

Chapter 19 SUMMARY

Outcomes

Knowledge

- summarize the historical events that led to the discovery of the structure of the DNA molecule, as demonstrated by Franklin, Watson, and Crick (19.3)
- explain the limitations of variability due to gene linkage and the influence of crossing over on assortment of genes on the same chromosome (19.2)
- explain the relationship between variability and the number of genes controlling a trait (19.2)
- compare the pattern of inheritance produced by genes on the sex chromosomes to that of genes on autosomes, as investigated by Morgan and others (19.1)

STS

- explain that decisions regarding the application of scientific and technological development involve a variety of perspectives including social, cultural, environmental, ethical, and economic considerations (19.2, 19.3)

Skills

- ask questions and plan investigations (19.2, 19.3)
- conduct investigations and gather and record data and information (19.1, 19.2)
- analyze data and apply mathematical and conceptual models by analyzing crossover data for a single pair of chromosomes to create a chromosome map showing gene arrangement and relative distance (19.2)
- work as members of a team and apply the skills and conventions of science (all)

Key Terms

19.1

autosome
linked genes
sex-linked trait
recessive lethal
Barr body

19.2

linkage group
locus (loci)
marker gene

19.3

continuity of life
bacteriophage
isotope
radioisotope
nucleotide
deoxyribose sugar
nitrogenous base
phosphate group

MAKE a summary

1. Create a poster of a human genome that shows the principles of sex-linked genes and helps show the relationship between genes and chromosomes. Label the sketch with as many of the key terms as possible. Check other posters and use appropriate ideas to make your poster clear.
2. Revisit your answers to the Starting Points questions at the beginning of the chapter. Would you answer the questions differently now? Why?

Go To www.science.nelson.com GO

The following components are available on the Nelson Web site. Follow the links for *Nelson Biology Alberta 20–30*.

- an interactive Self Quiz for Chapter 19
- additional Diploma Exam-style Review Questions
- Illustrated Glossary
- additional IB-related material

There is more information on the Web site wherever you see the Go icon in the chapter.

EXTENSION

CBC radioONE QUIRKS & QUARKS

Beyond the Genome

Dr. Victor Ambros (professor of genetics at Dartmouth Medical School), Dr. Katherine Wilson (associate professor of cell biology at Johns Hopkins Medical School), and Dr. Wolf Reik, (Babraham Institute in Cambridge, England) discuss their research on how our cells really work, including how genes "know" to turn on at the right times.

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Many of these questions are in the style of the Diploma Exam. You will find guidance for writing Diploma Exams in Appendix A5. Science Directing Words used in Diploma Exams are in bold type. Exam study tips and test-taking suggestions are on the Nelson Web site.

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DO NOT WRITE IN THIS TEXTBOOK.

Part 1

1. In performing experiments with fruit flies, *Drosophila melanogaster*, Thomas Morgan discovered that white eye colour is recessive to red eye colour. When females with white eyes were crossed with males with red eyes, Morgan discovered the females all had red eyes and the males all had white eyes. Select the answer that explains this outcome.

A. Male offspring inherit the white allele from the mother, which in males becomes dominant. Female offspring inherit the red allele from the father, which is dominant over the white allele they inherit from the mother.

B. Male offspring inherit the white allele from the mother and a Y chromosome from the father that does not carry a gene for eye colour. Female offspring inherit the red allele from the father, which is dominant over the white allele they inherit from the mother.

C. Male offspring inherit the red allele from the mother, which is recessive in males. Female offspring inherit the red allele from the father and no allele for eye colour from the mother.

D. Male offspring inherit the red allele from the father and a Y chromosome from the mother that carries an allele for white eye colour. Female offspring inherit the red allele from the mother, which is dominant over the white allele they inherit from the father.

Use the following information to answer questions 2 to 4.

In the pedigree chart shown in **Figure 1**, females are represented by circles and males by squares, while light shading indicates normal phenotype and dark shading indicates Duchenne muscular dystrophy.

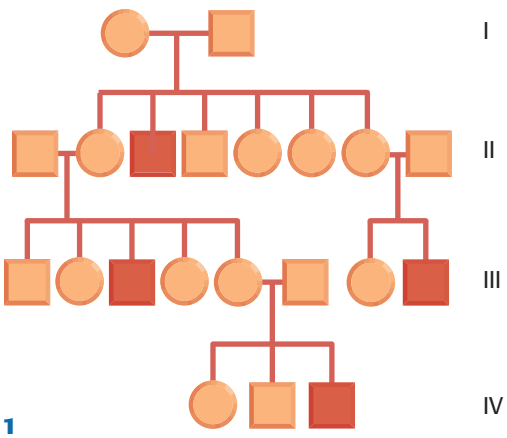

Figure 1

2. Identify the statement that correctly describes how Duchenne muscular dystrophy is inherited.

A. Duchenne muscular dystrophy is a dominant allele located on an autosome.

B. Duchenne muscular dystrophy is a recessive allele located on an autosome.

C. Duchenne muscular dystrophy is a dominant allele located on a sex chromosome.

D. Duchenne muscular dystrophy is a recessive allele located on a sex chromosome.

3. Identify the statement that is true for generation II.

A. 50 % of the males inherited the disorder from an allele carried by their mother.

B. 25 % of the males inherited the disorder from an allele carried by their mother.

C. 50 % of the females inherited the disorder from an allele carried by their father.

D. 100 % of the females inherited the allele carried by their mother but did not develop the disorder.

4. Identify the answer that is correct for generations I and III.

A. In generation I, the mother carries the recessive allele and is heterozygous. In generation III, males and females inherit the Duchenne allele from their mothers.

B. In generation I, the father carries the recessive allele and is heterozygous. In generation III, females and males inherit the Duchenne allele from their fathers.

C. In generation I, the mother carries the recessive allele and is homozygous. In generation III, only males inherit the Duchenne allele from their mothers.

D. In generation I, the father carries the recessive allele and is homozygous. In generation III, females and males inherit the Duchenne allele from their fathers.

5. Brown spotting on the teeth is a sex-linked trait in humans. A father with brown spotting passes the trait along to all his daughters but not to his sons. The mother does not have brown spotting on her teeth. This indicates that the brown spotting gene is

A. dominant and located on the X chromosome

B. recessive and located on the X chromosome

C. dominant and located on the Y chromosome

D. recessive and located on the Y chromosome

6. The recombination frequency among genes found on the same chromosomes depends on

A. which genes are dominant and which genes are recessive

B. the number of genes along the chromosome

C. the size of the chromosome

D. the distance between the genes

7. Ocular albinism in humans is characterized by a lack of pigment in the iris of the eyes. This X-linked trait often results in blindness for those afflicted. A woman who carries this trait marries a normal man. Identify the chance of ocular albinism in a child from this couple.
   A. 100 % chance of normal female offspring and a 100 % chance of normal male offspring
   B. 50 % chance of female offspring with ocular albinism, 50 % chance of normal female offspring, and 100 % chance of normal male offspring
   C. 100 % chance of normal female offspring, 50 % chance of male offspring with ocular albinism, and 50 % chance of normal male offspring
   D. 50 % chance of female offspring with ocular albinism, 50 % chance of normal female offspring, 50 % chance of male offspring with ocular albinism, and 50 % chance of normal male offspring

8. The allele *R* produces rose combs in chickens. Another allele *P,* located on a different chromosome, produces pea combs. The absence of the dominant rose comb and pea comb alleles (*rrpp*) produces birds with single combs. When the dominant *R* allele and the dominant *P* allele are both present, they interact to produce a walnut comb (*R_P_*). Identify the phenotypes of the parents and the expected phenotypic ratios of the F_1 generation from a cross of chickens with the genotype *RrPp* × *rrPp*.
   A. The parental phenotypes are walnut comb and pea comb. The expected F_1 phenotypic ratio from the cross is 3 walnut:3 pea:1 rose:1 single.
   B. The parental phenotypes are walnut comb and pea comb. The expected F_1 phenotypic ratio from the cross is 4 walnut:4 rose.
   C. The parental phenotypes are rose comb and pea comb. The expected F_1 phenotypic ratio from the cross is 3 walnut:2 rose:2 pea:1 single.
   D. The parental phenotypes are pea comb and single comb. The expected F_1 phenotypic ratio from the cross is 4 rose:4 pea.

Use the following information to answer questions 9 and 10.

The chromosome map in **Figure 2** shows the portion of a chromosome that carries genes for scalloped wings, bar eyes, and garnet eyes—all mutant traits in *Drosophila melanogaster*. It was drawn using data from several test crosses.

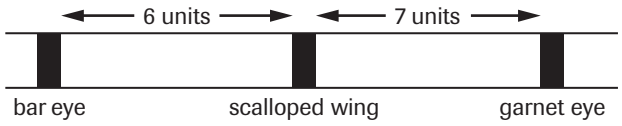

Figure 2

9. NR Determine the frequency of crossover between scalloped wings and garnet eyes, as a percent. (Record all four digits of your answer.)

10. NR Determine the frequency of crossover between bar eyes and garnet eyes, as a percent. (Record all four digits of your answer.)

Part 2

11. **Describe** Erwin Chargaff's contribution to the determination of DNA structure.

12. **Explain** how the development of the chromosome theory is linked with the development of the light microscope.

13. **Describe** the contributions made by Walter Sutton, Theodor Boveri, and Thomas Morgan in the development of the modern-day chromosome theory of genetics.

14. The gene for wild-type eye colour is dominant and sex-linked in *Drosophila melanogaster*. White eyes are recessive. The mating of a male with wild-type eye colour with a female of the same phenotype produces offspring that are $\frac{3}{4}$ wild-type eye colour and $\frac{1}{4}$ white-eyed. **Predict** the genotypes of the P_1 and F_1 generations.

15. The autosomal recessive allele *tra* transforms a female *Drosophila melanogaster* into a phenotypic male when it occurs in the homozygous condition. The transformed females are sterile. The *tra* gene has no effect on the phenotype of XY males. Using Punnett squares, **predict** the genotypes and phenotypes of individuals in the F_1 and F_2 generations from the following cross: XX, + /*tra* crossed with XY, *tra*/*tra*. (Note the + indicates the normal dominant gene.)

16. Edward Lambert, an Englishman, was born in 1717. Lambert had a skin disorder that was characterized by very thick skin, which was shed periodically. The hairs on his skin were very course and quill-like, giving him the name "porcupine man." Lambert had six sons, all of whom exhibited the same traits. The trait never appeared in his daughters. In fact, the trait has never been recorded in females. **Hypothesize** the nature of the inheritance of the "porcupine trait" that would explain these observations.

Use the following information to answer questions 17 to 20.

Figure 3 is a pedigree chart of a family in which some members have hemophilia.

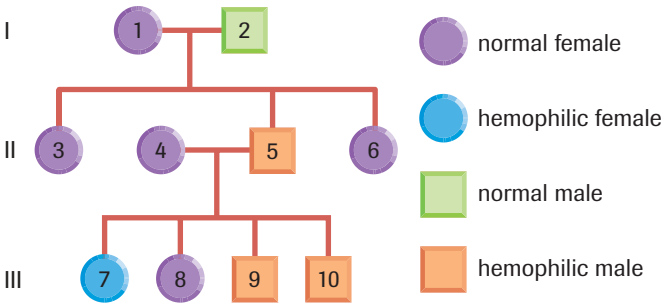

Figure 3

17. **Predict** the phenotypes of the P_1 generation.
DE

18. If parents 1 and 2 were to have a fourth child, **determine** the probability that the child would have hemophilia.
DE

19. If parents 1 and 2 were to have a second male, **determine** the probability that the boy would have hemophilia.
DE

20. **Predict** the genotypes of parents 4 and 5.
DE

21. A science student **hypothesizes** that dominant genes occur with greater frequency in human populations than recessive genes occur. Either support or refute the student's hypothesis, using the information that you have gathered in this chapter to **justify** your decision.

Use the following information to answer questions 22 to 24.

In 1911, Thomas Morgan collected the gene crossover frequencies shown in **Table 1** while studying *Drosophila melanogaster*. The loci for four different genes that code for wing shape are located on the same chromosome. Bar-shaped wings are indicated by the *B* allele, carnation wings by the *C* allele, fused veins on wings by the *FV* allele, and scalloped wings by the *S* allele.

Table 1

Gene combinations	Recombination frequency
FV/B	2.5 %
FV/C	3.0 %
B/C	5.5 %
B/S	5.5 %
FV/S	8.0 %
C/S	11.0 %

22. Use the crossover frequencies to **sketch** a gene map.
DE

23. **Identify** which genes are farthest apart. **Determine** their distance. **Illustrate** your answer by way of a diagram.
DE

24. From the data provided in **Table 1**, **conclude** in a written statement the relative position of the *FV*, *C*, and *B* alleles.
DE